



Key Actions Developed by the ERN Coordinators Group and the Networks

Franz Schaefer

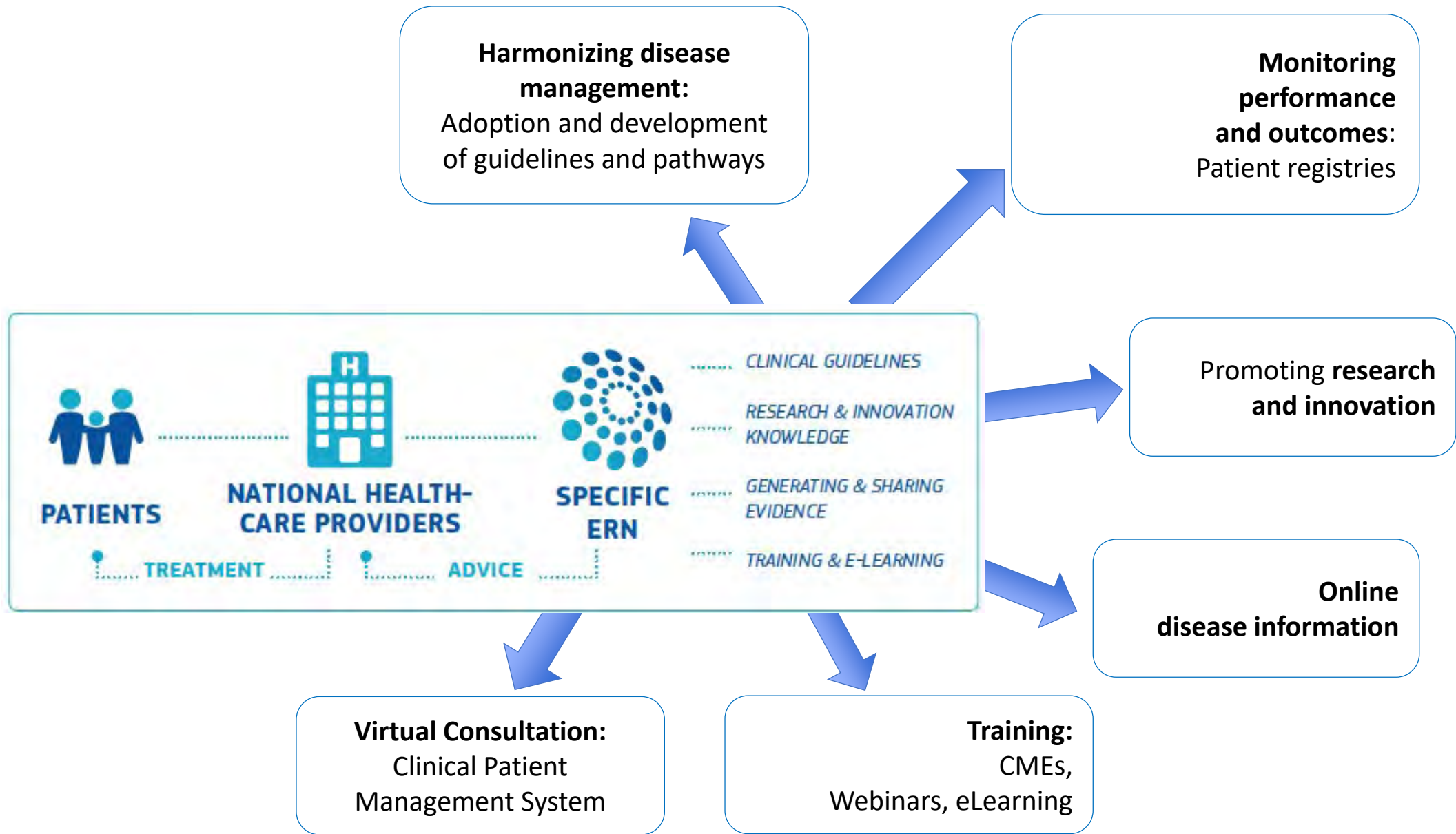
Heidelberg, Germany

Chair, ERN Coordinators Group

Coordinator, European Reference Network for Rare Kidney Diseases (ERKNet)



I have no disclosures to make with respect to the contents of this presentation.



Harmonizing disease management:
Adoption and development of guidelines and pathways

Monitoring performance and outcomes:
Patient registries

PATIENTS **NATIONAL HEALTH-CARE PROVIDERS** **SPECIFIC ERN**

TREATMENT **ADVICE**

..... *CLINICAL GUIDELINES*
..... *RESEARCH & INNOVATION KNOWLEDGE*
..... *GENERATING & SHARING EVIDENCE*
..... *TRAINING & E-LEARNING*

Promoting research and innovation

Online disease information

Virtual Consultation:
Clinical Patient Management System

Training:
CMEs, Webinars, eLearning

Review of Existing Evidence Base e.g. ReConnet

October 2018 - Volume 4 - Suppl 1

European Reference Networks: the first year activities of the ERN-ReCONNET on Clinical Practice Guidelines for rare and complex connective tissue and musculoskeletal diseases

AUTOIMMUNITY

REVIEW

[Review: Antiphospholipid syndrome: state of the art on clinical practice guidelines \(18 October, 2018\)](#) 

Marteen Limper, Carlo Alberto Scirè, Rosaria Talarico, Zahir Amoura, Tadej Avcin, Martina Basile, Gerd Burmester, Linda Carli, Ricard Cervera, Nathalie Costedoat-Chalumeau, Andrea Doria, Thomas Dörner, João Eurico Fonseca, Ilaria Galetti, Eric Hachulla, David Launay, Filipa Lourenco, Carla Macieira, Pierluigi Meroni, Carlo Maurizio Montecucco, Maria Francisca Moraes-Fontes, Luc Mouthon, Cecilia Nalli, Yeronique Ramoni, Maria Tektonidou, Jacob M van Laar, Stefano Bombardieri, Matthias Schneider, Vanessa Smith, Ana Vieira, Maurizio Cutolo, Marta Mosca, Angela Tincani

CONNECTIVE TISSUE DISEASES

REVIEW

[Review: Rare diseases under different levels of economic analysis: current activities, challenges and perspectives \(12 November, 2018\)](#) 


Sara Cannizzo, Valentina Lorenzoni, Ilaria Palla, Salvatore Pirri, Leopoldo Trieste, Isotta Triulzi, Giuseppe Turchetti

[Review: Ehlers-Danlos syndromes: state of the art on clinical practice guidelines \(18 October, 2018\)](#) 

Alberto Sulli, Rosaria Talarico, Carlo Alberto Scirè, Tadej Avcin, Marco Castori, Alessandro Ferraris, Charissa Frank, Jürgen Grunert, Sabrina Paolino, Stefano Bombardieri, Matthias Schneider, Vanessa Smith, Maurizio Cutolo, Marta Mosca, Fransiska Malfait

[Review: Relapsing polychondritis: state of the art on clinical practice guidelines \(18 October, 2018\)](#) 

Simona Rednic, Laura Damian, Rosaria Talarico, Carlo Alberto Scirè, Alexander Tobias, Nathalie Costedoat-Chalumeau, David Launay, Alexis Mathian, Lisa Matthews, Cristina Ponte, Paola Toniati, Stefano Bombardieri, Charissa Frank, Matthias Schneider, Vanessa Smith, Maurizio Cutolo, Marta Mosca, Laurent Arnaud

[Review: Mixed connective tissue disease: state of the art on clinical practice guidelines \(18 October, 2018\)](#) 

Review of Existing Evidence Base

e.g. ReConnet

Evaluation and endorsement of existing guidelines

e.g. ERKNet

Thematic Area: **Alport Syndrome**

The following guidance documents have been adopted based on standardized reviews and are followed in all ERKNet centers:

- 1) **Expert Guidelines for the Management of Alport Syndrome and Thin Basement Membrane Nephropathy.**
J Am Soc Nephrol 2013; 24:364-75

Core Recommendations:

1. Genetic testing is the gold standard for the diagnosis of Alport syndrome and the demonstration of its mode of inheritance.
2. All affected members of a family with X-linked Alport syndrome, including most mothers of affected males, should be identified and followed.
3. Males with X-linked Alport syndrome and individuals with autosomal recessive disease should be treated with renin-angiotensin system blockade.
4. Affected mothers of males with X-linked Alport syndrome should be discouraged from renal donation because of their own risk of kidney failure.
5. Genetic testing should be considered in some individuals with thin basement membrane nephropathy to exclude X-linked Alport syndrome.

Comments by evaluators:

- Useful and comprehensive guidelines.
- The recommendations are described as expert opinions (Level D) / opinions of respected authorities (Level III)
- The health questions are specifically described and further categorised by sub types- Alport syndrome (X-Linked, autosomal recessive) and thin basement membrane nephropathy.
- The term autosomal dominant Alport syndrome is not discussed in these guidelines.

- 2) **Clinical practice recommendations for the treatment of Alport syndrome:**
A statement of the Alport Syndrome Research Collaborative.
Pediatr Nephrol 2013; 28:5-11

Review of Existing Evidence Base

e.g. ReConnet...

Evaluation and endorsement of existing guidelines

e.g. ERKNet...

New Guideline Development

e.g. Euracan/PaedCan, ERN-Ithaca...

- Collaboration with professional societies
- Collaborations between ERNs
- ERN Taxonomy Project

CLINICAL PRACTICE GUIDELINES

Bone sarcomas: ESMO–PaedCan–EURACAN Clinical Practice Guidelines for diagnosis, treatment and follow-up[†]

P. G. Casali^{#1}, S. Bielack^{#2}, N. Abecassis³, H.T. Aro⁴, S. Bauer⁵, R. Biagini⁶, S. Bonvalot⁷, I. Boukovinas⁸, J. V. M. G. Bovee⁹, B. Brennan¹⁰, T. Brodowicz¹¹, J. M. Broto¹², L. Brugières¹³, A. Buonadonna¹⁴, E. De Álava¹⁵, A. P. Dei Tos¹⁶, X. G. Del Muro¹⁷, P. Dileo¹⁸, C. Dhooge¹⁹, M. Eriksson²⁰, F. Fagioli²¹, A. Fedenko²², V. Ferraresi⁶, A. Ferrari²³, S. Ferrari²⁴, A. M. Frezza²⁵, N. Gaspar¹³, S. Gasperoni²⁶, H. Gelderblom²⁷, T. Gil²⁸, G. Grignani²⁹, A. Gronchi¹, R. L. Haas³⁰, B. Hassan³¹, S. Hecker-Nolting², P. Hohenberger³², R. Issels³³, H. Joensuu³⁴, R. L. Jones³⁵, I. Judson³⁶, P. Jutte³⁷, S. Kaal³⁸, L. Kager³⁹, B. Kasper³², K. Kopeckova⁴⁰, D. A. Krákorová⁴¹, R. Ladenstein³⁹, A. Le Cesne¹³, I. Lugowska⁴², O. Merimsky⁴³, M. Montemurro⁴⁴, B. Morland⁴⁵, M. A. Pantaleo⁴⁶, R. Piana²¹, P. Picci²⁴, S. Piperno-Neumann⁷, A. L. Pousa⁴⁷, P. Reichardt⁴⁸, M. H. Robinson⁴⁹, P. Rutkowski⁴², A. A. Safwat⁵⁰, P. Schöffski⁵¹, S. Sleijfer⁵², S. Stacchiotti²⁵, S. J. Strauss¹⁸, K. Sundby Hall⁵³, M. Unk⁵⁴, F. Van Coevorden⁵⁵, W.T.A. van der Graaf^{35,38,55}, J. Whelan¹⁸, E. Wardelmann⁵⁶, O. Zaikova⁵⁷ & J. Y. Blay⁵⁸, on behalf of the ESMO Guidelines Committee, PaedCan and ERN EURACAN*

Review of Existing Evidence Base

e.g. ReConnet...

Evaluation and endorsement of existing guidelines

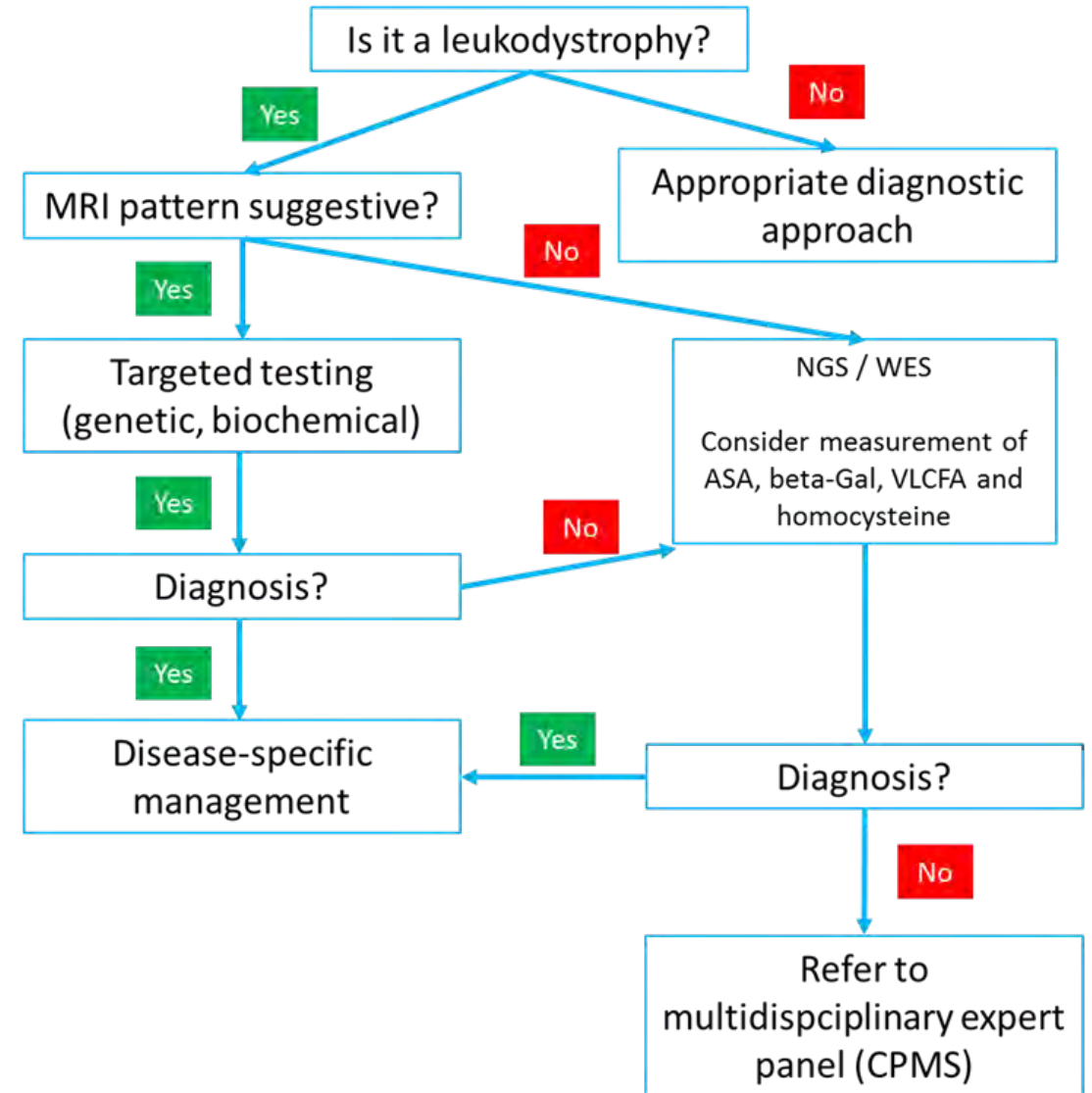
e.g. ERKNet...

New Guideline Development

e.g. Euracan/PaedCan, ERN-Ithaca...

Diagnostic Flowcharts, Clinical Rating Scales

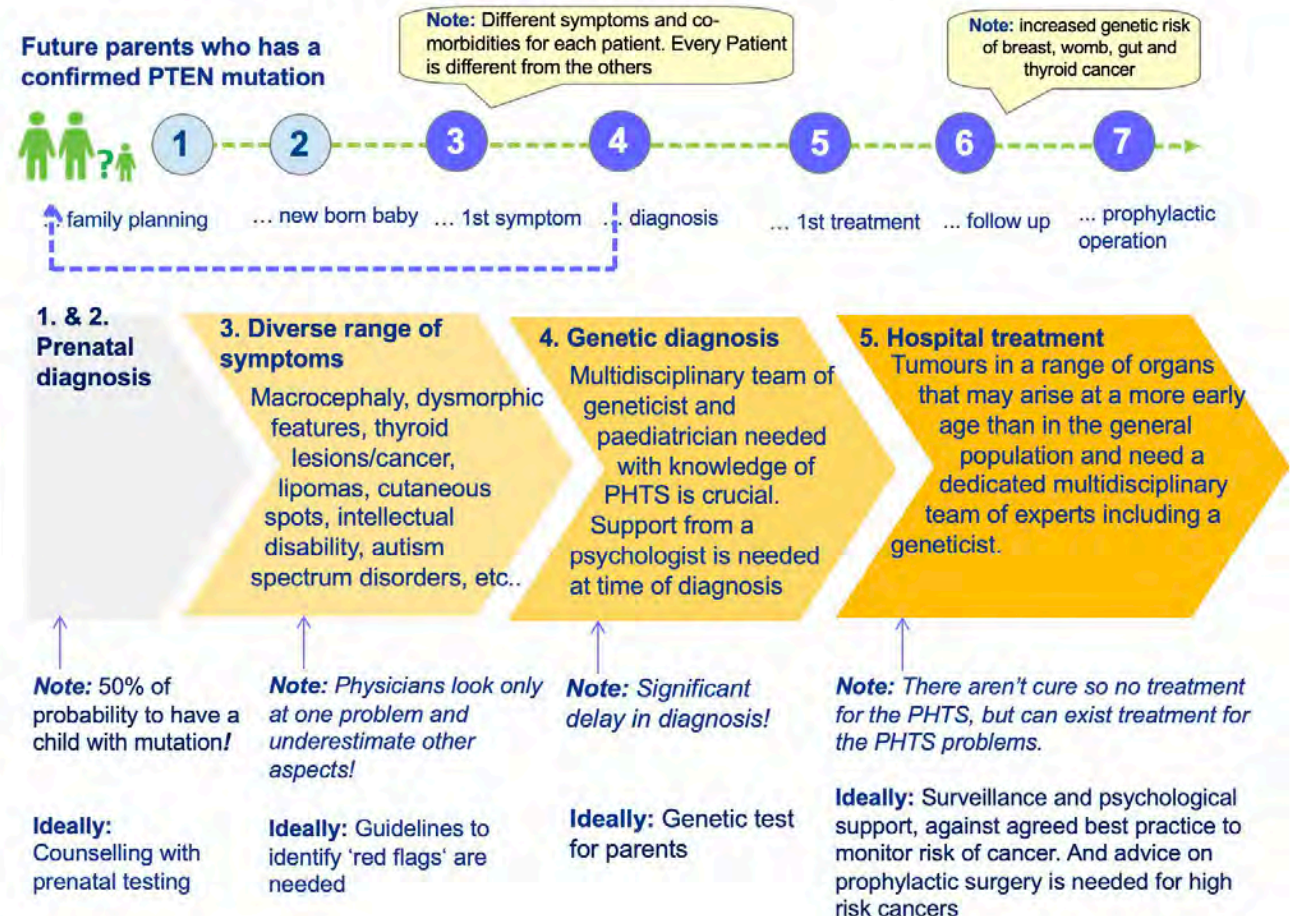
e.g. ERN-RND...



ERN Genturis: “Patient Journeys”

- Developed by patients
- Medical details approved by ERN professionals
- Contents:
 - Typical patient pathway
 - Typical challenges and pitfalls
 - Medical issues where specialists can help
 - Unmet patient needs at various disease stages

Patient Journey PTEN hamartoma tumour syndrome (PHTS)



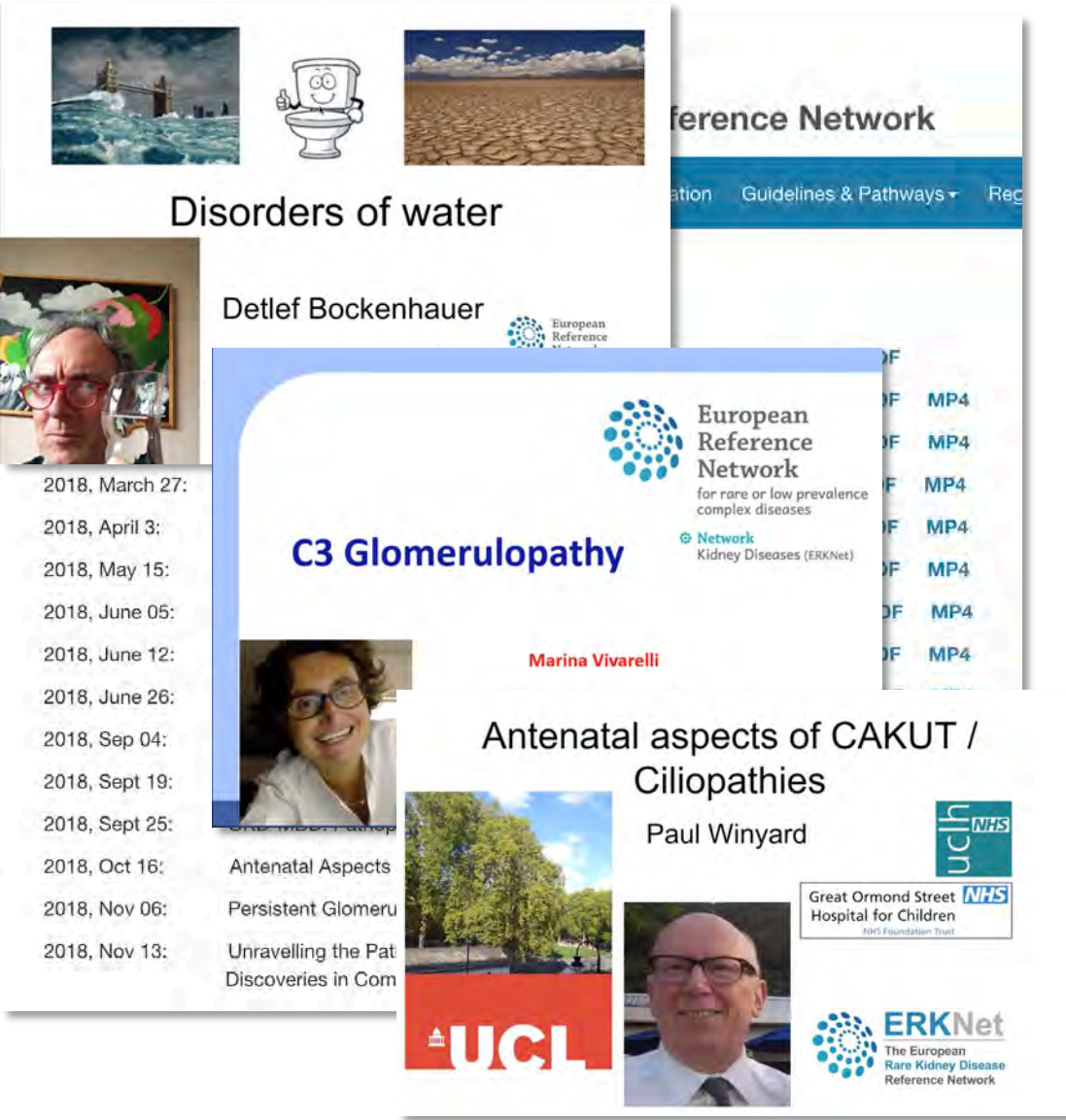
All ERNs organizing CME Courses, summer/winter schools
- good collaborations with professional specialist societies

Webinars programs starting (e.g. Ithaca, ERKNet...)

CEF grant 2019/2020: **eLearning** platforms

ERN-RND, ERN-Eye: Clinical fellowship exchange grants

-> Funding for regular **clinical exchange training programs needed**



Disorders of water

Detlef Bockenhauer

C3 Glomerulopathy

Marina Vivarelli

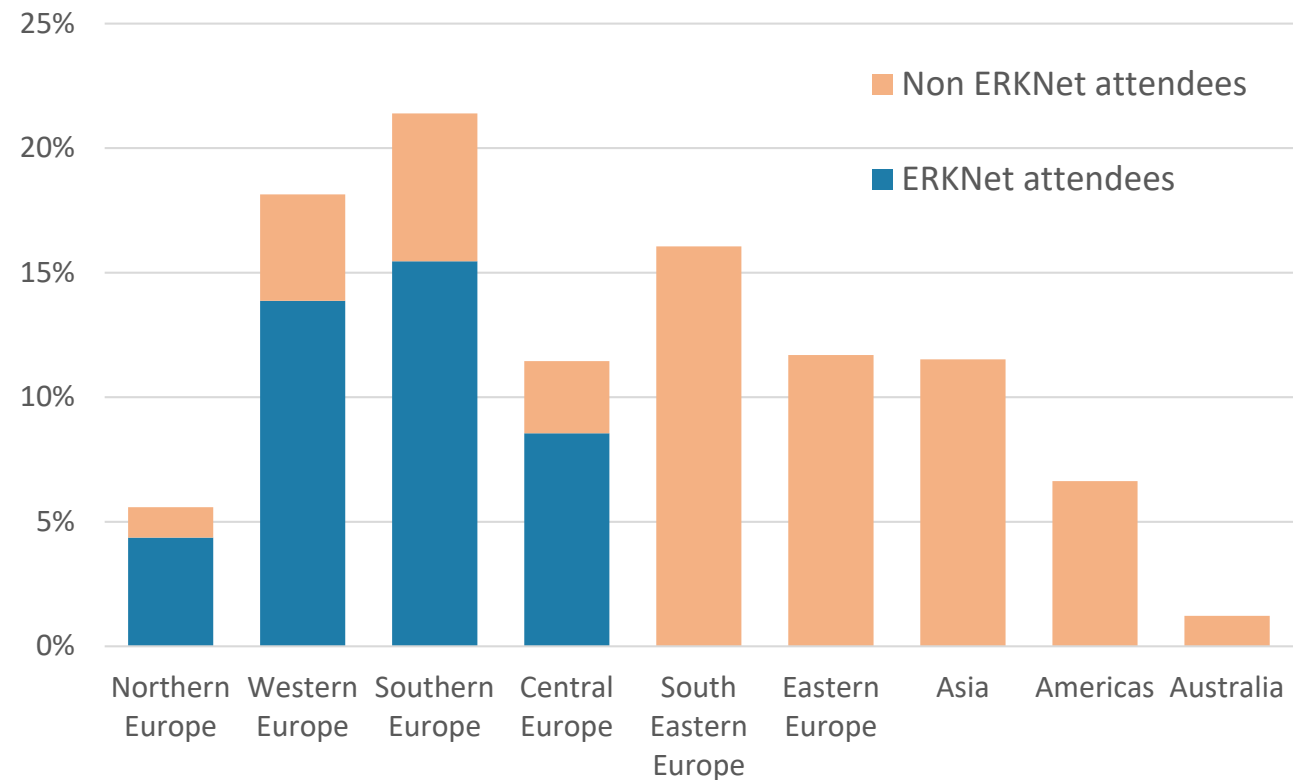
Antenatal aspects of CAKUT / Ciliopathies

Paul Winyard

2018, March 27:
 2018, April 3:
 2018, May 15:
 2018, June 05:
 2018, June 12:
 2018, June 26:
 2018, Sep 04:
 2018, Sept 19:
 2018, Sept 25:
 2018, Oct 16:
 2018, Nov 06:
 2018, Nov 13:

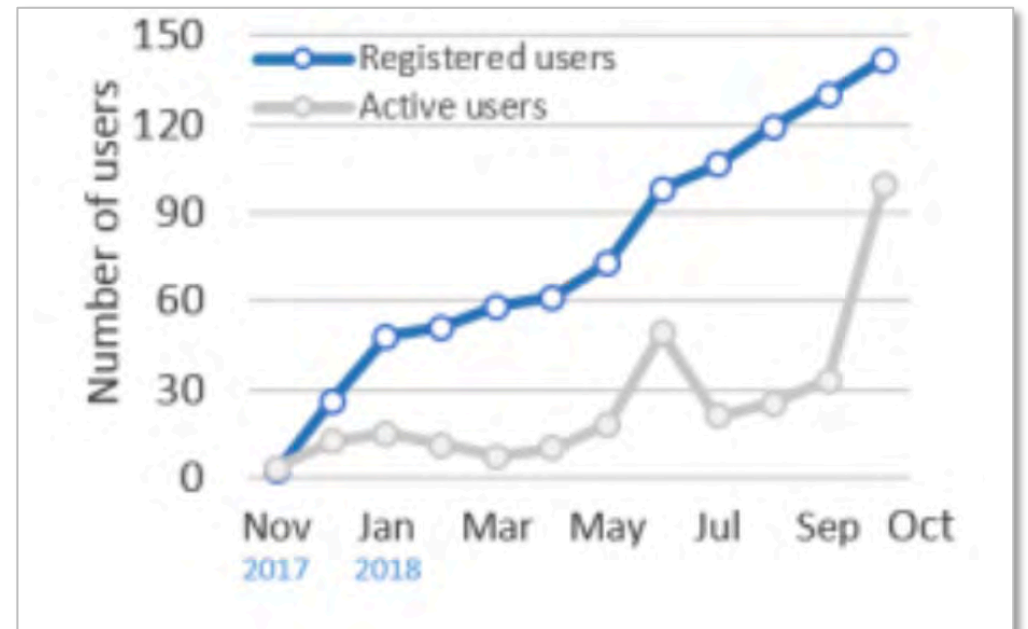
Antenatal Aspects
 Persistent Glomeru
 Unravelling the Pat
 Discoveries in Com

Biweekly live streams
 Announced via ERN & professional society newsletters
 100-150 registrations per Webinar
 50-100 live attendees, 150-200 post-event downloads



Early experience:

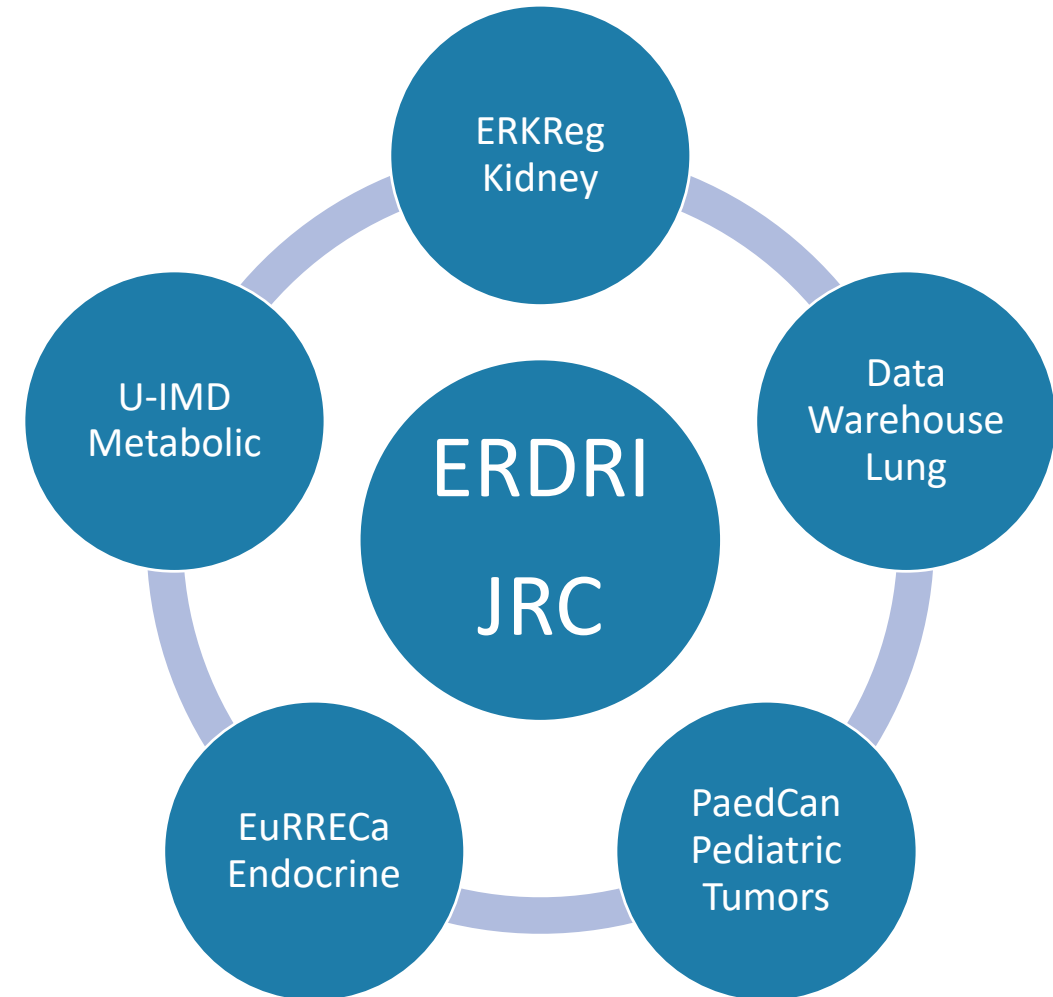
- eConsulting platform with unique features:
Advanced image/histopathology slide viewing functionalities
- Potentially cost-effective tool by sparing physical cross-border patient referral
- Time-consuming data entry,
difficulty to arrange multi-expert VCs
- Operational help desks being implemented
- Data protection issues by GDPR
- Re-think target groups / access concept :
„Experts rarely need expert advice“



Keys to success:

- Definition of use indications and (national) access pathways
- Awareness of medical community
- Streamlined / customized data entry
- In the long run: Reimbursement system for expert services

- High priority for most ERNs
- Health Programme Research Call:
Registries for European Reference Networks
- 5 ERNs funded to develop core registries
- Collaboration with JRC
European RD Registration Platform
- Further EU support envisioned
- Local center support will be key to success



ERN Research Working Group:

- **Mapping** ERN research capacities ongoing
- **Strategy Workshop** Jan 24 2019

Pediatric clinical research:

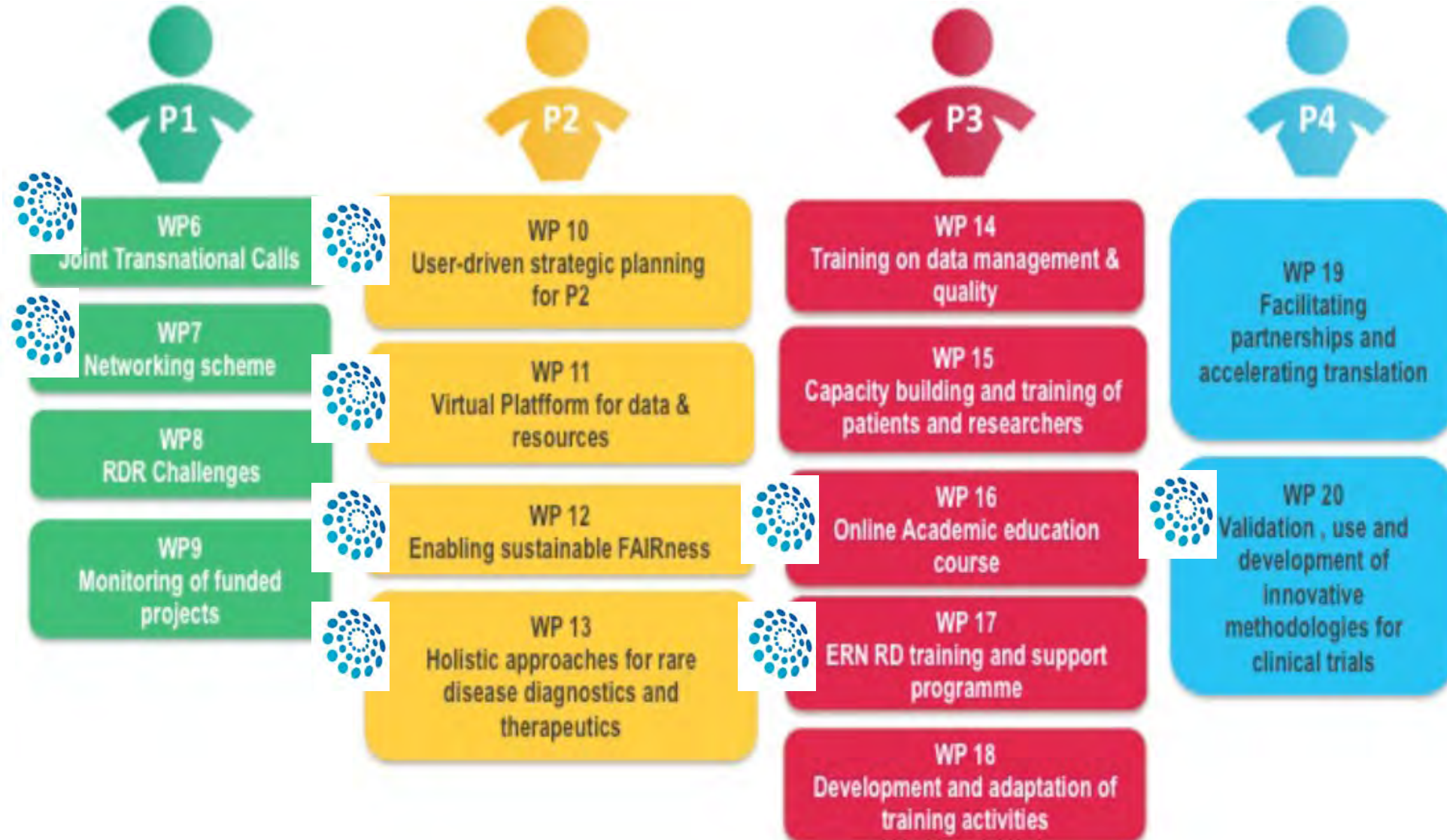
Involvement in **conect4children** Paneuropean Clinical Research Network

Transveral COST proposal:

EaRN-Gen for promotion of genetic diagnostics

European Joint Programme for Rare Disease Research (**EJP-RD**):

Multi-level involvement of ERNs



Dissemination Activities



Following 171 Followers 345 Likes 102 Lists 0 Moments 0

Tweets Tweets & replies Media

ERKNet @EuRefNetwork · 18h
HDF, Heart and Height (3H) Study is the largest study to date led by dr. Shroff from @GreatOrmondSt that investigates outcomes of #HDF vs conventional #HD in children

BMC Nephrology
Effect of haemodiafiltration vs conventional h...
Cardiovascular disease is prevalent in children on dialysis and accounts for almost 30% of all deaths. Randomised trials in adults suggest that haemo...
bmcnephrol.biomedcentral.com

ERKNet @EuRefNetwork · Oct 2
The utilization of small #pediatric #donors #kidneys for #transplantation reviewed in @Ped_Neph

Utilisation of small paediatric donor kidneys f...
With the increasing need for kidney transplantation in the paediatric population and changing donor demographics, children without a living donor opt...
link.springer.com

ERKNet @EuRefNetwork · Sep 30
#CD8 T-cells modulate #ADPKD progression

CD8+ T cells modulate autosomal dominant p...
Autosomal dominant polycystic kidney disease (ADPKD) is the most prevalent inherited nephropathy. To date, therapies alleviating the d...
kidney-international.org

ERKNet @EuRefNetwork · Sep 29
An update on #LDL #apheresis in #nephrotic syndrome @Ped_Neph

An update on LDL apheresis for nephrotic syn...
Low-density lipoprotein (LDL) apheresis has been used increasingly in clinical practice for the treatment of renal diseases with nephrotic synd

European Reference Network
for rare or low prevalence complex diseases

Network
Vascular Diseases (VASCERN)

Newsletter #23 - October 2018

View this email online

VASCERN Days 2018: Highlights from our 2nd Annual Meeting
VASCERN's second annual seminar place on October 11-12th, 2018 in Paris, France! In attendance were 70 of our 31 Healthcare Provider Members as well as European Patient Ad

VASCERN Representation in the interERN WG on CPMS
VASCERN co-announce the

European Reference Network
for rare or low prevalence complex diseases

Network
Vascular Diseases (VASCERN)

Thursday 18 October 2018

ERN

GUARD-Heart
Gateway to Uncommon And Rare Diseases of the Heart

ERIN GUARD-HEART SPECIAL NEWSLETTER

YEAR 2018 NUMBER 6

ERN GUARD-Heart CPMS Helpdesk
Amsterdam, 2 October 2018

Since 1 September 2018 the ERN GUARD-Heart CPMS helpdesk is open! The CPMS Helpdesk will facilitate and ensure adequate and efficient use of the CPMS and the IT Collaborative Platform within ERN GUARD-Heart.

We will be trained by the ERN support team so we will be equipped with the right knowledge to support all ERN GUARD-Heart members. We will train the ERN GUARD-Heart members to use CPMS and the IT Collaborative Platform, assist the ERN members in adding patient data in CPMS and create panels, provide support for local hardware and/or software, provide phone support and if necessary face-to-face support for all ERN GUARD-Heart members.

The CPMS helpdesk is situated in the Amsterdam Medical Center. If you have any questions regarding CPMS or the IT Collaborative Platform, please let us know by sending an email to cpmshelpdesk@guardheart.ern-net.eu. Or you can call to +31 6 45153319 from 9:00 to 17:00 CET from Monday to Friday. If necessary we can schedule a call outside office hours.

We hope to hear from all of you!

ERN GUARD-Heart CPMS Helpdesk
Phone: +31 6 45153319
Mail: cpmshelpdesk@guardheart.ern-net.eu

European Reference Networks
Share. Care. Cure.

Endo-ERN
European Reference Network on Rare Endocrine Conditions

Newsletter October

European Reference Networks
FOR RARE, LOW-PREVALENCE AND COMPLEX DISEASES

Share. Care. Cure.

European Commission | Health

Translational Summer School Special 2018!!

Neuromuscular Translational Summer School
2-6 July 2018

TREAT-NMD

Our translational summer school is fast approaching and we have a few places available for those who are interested in attending. This month's newsletter provides a comprehensive overview of the programme, highlighting the key stakeholders as well as the objectives of each session.

The course, which is run in association with TREAT-NMD, costs €1000 and will be held on 2-6 July in Newcastle-upon-Tyne, UK.

Attendees will benefit from a comprehensive grounding in the translational research

European Reference Network
for rare or low prevalence complex diseases

Network
Neuromuscular Disorders (ERN NMD)

ERN-EYE
THE EUROPEAN REFERENCE NETWORK DEDICATED TO RARE EYE DISEASES

13 founding members

29 hospitals

900 rare eye patients

24 000 genetic P. M. cases

Watch the new ERN-EYE video!

ERNs - Meeting of the coordinators
All coordinators of the ERNs met on 26th June in Brussels for the coordinators meeting. They discussed about the accomplishments and challenges of the ERNs with the European Commission.

CPMS - Customization of the platform
The Clinical Patient Management System (CPMS) was launched by the European Commission (DG SANTE) Monday 20th November, 2017. It is a web-based application where healthcare professionals from the European Reference Networks (ERNs) will be able to discuss real patient cases.

Expected ERN Expansion: The Coordinators' View

Geographic expansion and **closure of expertise gaps** will be vital to materialize full potential of ERNs

Full membership and Affiliated Partnerships are equally valid models of cooperation

While new partners are welcome, **careful selection** according to competency criteria and ERN needs will be essential to

- **maintain quality** and
- **avoid operational challenges**
(in view of unchanged coordination budgets)

