

Endo-ERN Steering Committee / Advisory Board meeting

Date: Tuesday 20 January 2026

Time: 09:30 – 11:30 hrs



Funded by
the European Union

IN MEMORIAM



Jette Kristensen



Juliane Leger



ADDISON FORENINGEN I DANMARK

Agenda

1. Review (outstanding) action points previous meeting
2. Work Package updates (8 minutes each)
3. Working Groups updates (8 minutes each)
4. Other RD project updates
5. AOB & questions

Review outstanding action points previous SCAB meeting

1. Pre-plan MTG-specific HCP meetings 2026

MTG	Date	Time
1	To be scheduled	
2	18 March 2026	16:00 – 17:00
3	25 February 2026	16:00 – 17:00
4	Next meeting @ GA	
5	26 January 2026	15:00 – 16:00
6	20 February 2026	12:00 – 13:00
7	4 February 2026 2 September 2026	16:00 – 17:00
8	15 October 2026 (TBC)	

2. Pre-plan minimum 5 webinars for early 2026

WP1 Management & Coordination

Alberto Pereira, Adult Chair Endo-ERN

Olaf Hiort, Paediatric Chair Endo-ERN

Charlotte van Beuzekom, Project Manager Endo-ERN



New ePAG MTG1 Adrenal

Teresa Valencia

Spanish National Association of Addison's Disease and other
Rare Endocrine Disorders





ESE and ESPE RD Committees



ESE RD Committee

<u>(co)Chairs</u>	<u>Term</u>
Elena Valassi (ESE)	2024 – 2028
Alberto Pereira (Endo-ERN)	2019 – 2027

Endo-ERN representatives

Gudmundur Johannsson	2020 – 2026
Marek Niedziela	2024 – 2028
Luca Persani	2024 – 2028
Dimitra Vassiliadi	2024 – 2028

Alignment procedures committee composition



ESPE RD Advisory Committee

<u>(co)Chairs</u>
Evangelia Charmandari
Marek Niedziela

Endo-ERN representatives

Olaf Hiort
Nicole Reisch (?)
Pietro Maffei

Funding grants



Current EU4Health grant 2023-2027

- Deliverables
 - All deliverable reports submitted in time
 - Under evaluation by EC and external evaluators
- 1st Period report
 - Covering 1 Oct '23 – 31 Dec '25
 - Technical and financial report
 - Grant partners informed about obligations/deadlines
 - Submission deadline total report to EC = end February '25

Future ERN grants

- Multi-annual Financial Framework 2028-2034
- High Level Meeting - Declaration

Submitted deliverables – due Nov 2025

Nr.	Name
D1.4	Organisation General Assembly Y2
D2.2	Dissemination report 1
D3.2	Evaluation report 1
D3.5	Indicators report Y2
D4.1	CPMS activities report Y1+Y2
D5.1	Registry activities report Y1+Y2
D6.1	Training activities report Y1+Y2
D7.1	CPG/CDST activities report Y1+Y2
D7.3	First new CPG produced
D8.1	Report activities WP8
D9.1	Report activities WP9

Endocrine Connections - special issue 2025



linkedin.com/company/rareendoern

linkedin.com/showcase/endocrineconnections

https://ec.bioscientifica.com

ENDO-ERN Vol II: Rare Diseases and Transition in Care

SPECIAL COLLECTION

IN COLLABORATION WITH:

Scan to learn more

Lead author	Paper title	Status
Felix Reschke	Challenges and Strategies for Transitioning Adolescents with Rare Forms of Diabetes to Adult Care: Perspectives and Evidence-Based Approaches	ACCEPTED
Nienke Biermasz	Quality of care for patients with prolactinoma diagnosed under the age of 25 years	Under review
Hedi Claahsen	Enhancing Transition Care for adolescents and young adults with adrenal insufficiency in the Netherlands: A Holistic Model for Improved Patient Outcomes	ACCEPTED
Corinna Grasemann	Continuity of Care in Klinefelter Syndrome: Age-Adapted Modules for Standardized Clinical Data Collection (I-KS)	ACCEPTED
Violeta Iotova	Feasibility of transition research in pituitary disease using patient registries	ACCEPTED
Ana Priego	The EuRECa Core Registry and its potential for studying transition of care in Endo-ERN reference centres	Waiting for submission
Emily White	First 5 Years of Endo-ERN: Evaluation of the Network and Member Expert Centres & Creation of Endo-ERN Performance System	Under review
	Acromegaly: The Patient Journey	Waiting for submission
Judit Toke	Transitional care of patients with congenital adrenal hyperplasia - our experiences and challenges in a tertiary adult referral center in Hungary	Under review
George Mastorakos	TRANSITION of pediatric patients on GH treatment in Greece	Waiting for submission
Andrea Isidori	Mapping Transition of Care for Rare Endocrine Conditions: Findings from a Cross-Sectional Survey by the Endo-ERN ToC Working Group	Under review
Violeta Iotova	Editorial	Waiting for submission

Endocrine Oncology - special issue 2026/27



General

- More clinical focused (relatively younger) journal
- Waiting for first impact factor, promising numbers

Topics of interest

- Gonadal diagnoses
- Side effects of immunotherapy
- Endocrine consequences of childhood cancer (cross-ERN collaboration with Paedcan?)
- Ultrarare: carney complex, aggressive pituitary tumors
- Cross-collaboration with other cancer-ERNs like EURACAN?

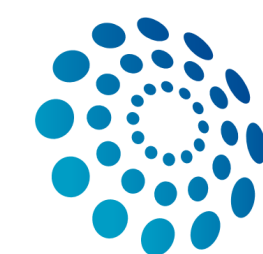
Proposal

- 8-10 papers (1 per MTG, 1 per WG)
- Free of charge, open access
- 2-4 guest editors

Next steps

- Coordination office sends invitation to all members
- MTG selection by MTG chairs
- Launch portfolio of possible papers during GA Athens





European
Reference
Network



Endo-ERN

Upcoming Meetings 2026

General Assembly 20-21 April, Athens, Greece

Monday 20 April

09:00 – 10:00 SCAB meeting

10:30 – 17:30 Plenary program

19:30 – 21:30 Dinner with Acropolis view

Tuesday 21 April

09:00 – 14:00 Plenary program

Closing lunch

Please make sure to book your travel/hotel and dinner ticket!

ECE2026 9-12 May, Prague, Czechia

Endo-ERN symposium 12 May & booth

- Introduction on Endo-ERN initiatives (focus EndoCompass paper) - *Alberto Pereira/Olaf Hiort*
- WG Genomic Testing - *Thomas Eggermann*
- The pediatric thyroid cancer registry – *Hanneke Van Santen*, UMC Utrecht, NL
- The reduced life expectancy and treatment options for RTHbeta – *Irene Campi*, I.R.C.C.S. Istituto Auxologico Italiano Milan, Italy
- ePAG presentation on patient journeys (*TBD*)

ESPE 2026 8-10 September, Marseille, France

Endo-ERN symposium & booth

WP2 Communication & Dissemination

Aimee Casey, Communication Manager, Endo-ERN



Funded by
the European Union



Social media update
January 2026

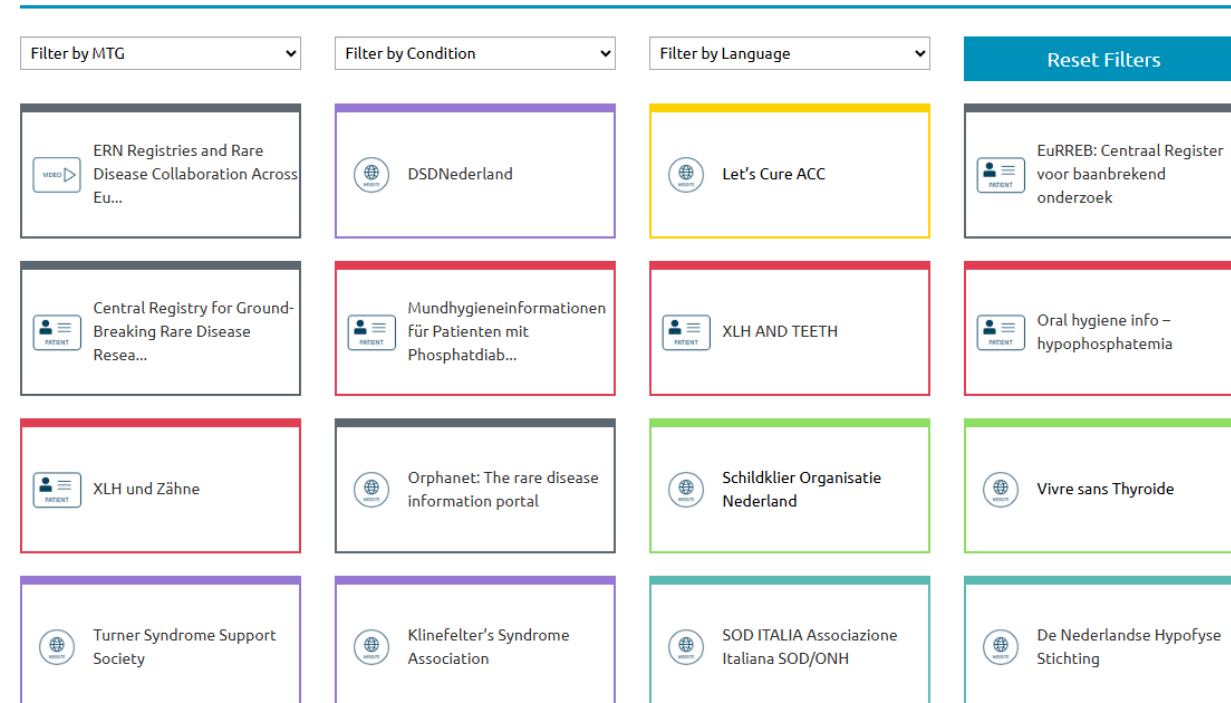
	Current total	Increase from September
LinkedIn	978	+246
Facebook	490	+34
YouTube	743	+99
Most popular newsletter	Clinical practice	



Patient Resources Process

- Materials sent to project office via online form
- Short summary in English describing the content and purpose of the materials
 - ePAG submitted materials should indicate if any medical information is included.
- The MTG chairs will review the submission and decide whether to accept and reject it based on the summary.

Patient Resources



We will include the following disclaimer: *This resource was contributed by Endo-ERN member name in language. The relevance of the topic has been reviewed by the appropriate Main Thematic Group (MTG). English summaries and translations into other languages were produced with the assistance of AI. Local-language versions have not been independently validated by Endo-ERN.*

New Patient Resources

- 20 plus Bulgarian patient materials for MTG3, MTG5, and MTG6
- MEN-1 layperson guideline (English, German, Spanish, Italian, Dutch)
- Recommended medical check-ups and examinations in XLH (English& German)

Patient Resources

Filter by MTG Filter by Condition Filter by Language [Reset Filters](#)

ERN Registries and Rare Disease Collaboration Across Eu...	DSDNederland	Let's Cure ACC	EuRREB: Centraal Register voor baanbrekend onderzoek
Central Registry for Ground-Breaking Rare Disease Resea...	Mundhygieneinformationen für Patienten mit Phosphatdiab...	XLH AND TEETH	Oral hygiene info – hypophosphatemia
XLH und Zähne	Orphanet: The rare disease information portal	Schildklier Organisatie Nederland	Vivre sans Thyroïde
Turner Syndrome Support Society	Klinefelter's Syndrome Association	SOD ITALIA Associazione Italiana SOD/ONH	De Nederlandse Hypofyse Stichting

Rare disease day 2026

- JARDIN campaign – patient focus
- Social posting
- Newsletter
- Downloadable poster – internal communication
- Text for internal communication - intranet or newsletter

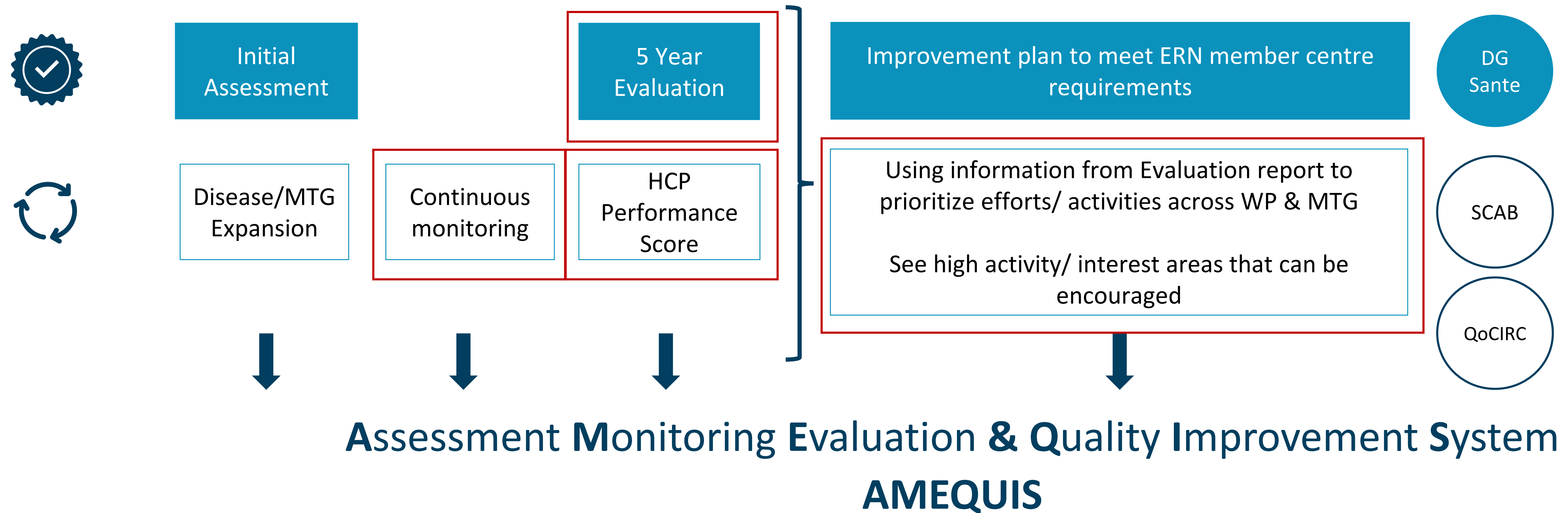


WP3 Evaluation

Emily White, Project Manager & Endo-ERN Operational Helpdesk Manager



Evaluation in Endo-ERN – QI



Continuous Monitoring

2025 data collection started from January 1st 2026

Key reminders to all members:



- Update your e-REC prior to January 31st
- Request reports of total number of patients in time (if necessary)

New data requested:

- Number of rare endocrine patients received/ referred from other EU countries in 2025 in your HCP
- Number of Ukrainian patients referred to or treated for rare endocrine conditions in your HCP in 2025
- Emergency/ potential crisis situations

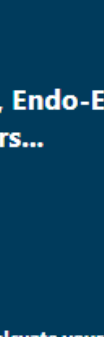
Data no longer requested

- Accredited educational activities run by your HCP – only those with ERN direct involvement are being gathered
- Non-accredited educational activities and all details therein

Endo-ERN Performance Report 2024

This report is generated within the Endo-ERN HCP Performance system. This mandatory evaluation exercise assesses the activity level of each of our member HCPs based on information from sources shown here.



In 2024, Endo-ERN members...

- Seen 21,074 confirmed new cases of rare endocrine patients
- Researcher with over 110 active clinical trials and studies
- Completed/launched over 350 rare disease specific training and educational activities
- Used Endo-ERN guidelines/EBPGs produced by Endo-ERN and European rare experts/Endocrine Council
- Attended and gave input to 121 congresses, conferences & meetings discussing rare endocrine disease/care
- Worked alongside experts from over 42 countries worldwide


How do we calculate your score?

There are 34 individual activity category.

Possible Points
21 pts **mandatory** = 60% of total
20 pts **expected** = 25% of total
22 pts **bonus** = 15% of total

Mandatory Activities = each HCP has to complete to retain membership (max. 3 points per activity)

Disclaimer: The scores generated within this activities as a member of our network (nationally or locally).




European Reference Network

for rare or low prevalence complex diseases

Network Endocrine Conditions (Endo-ERN)

Appendix 1 – Continuous Monitoring Indicators



KPI/Indicator	CM Indicator	No. reported
3.2a	Urgent MDT advice - Time in days (Referral-Panel closed in CPMS)	149 days
	Non-urgent MDT advice - Time in days (Referral-Panel open in CPMS)	



Funded by the European Union

3.2b	Panel closed in CPMS)	115 days	4 clinical exchange, 16 webinars, 2 endorsed events (CME request pending), 3 nationally accredited in host country.
4.1a	Number of non-accredited education/training activities developed and delivered by the coordination team or by members of the ERN	25	273 additional activities reported by members that were rare disease focused/ relevant.
4.1b	Number of trainees who completed non-accredited training	534	4 from exchange, 530 from webinars, other activities attendees not known
4.1c	Number of countries of origin of trainees who completed non-accredited training	43 countries	Approximation as some HCPs reported a world wide audience
			5 accredited webinar arranged in 2024 by Endo-ERN coordination . 1 endorsed activities run by members of ERN with CME credits approved.
4.2a	Number of accredited education/training activities developed and delivered by the coordination team OR by members of the ERN	6	288 activities reported by members that were rare disease focused/ relevant or had national accreditation only.
4.2b	Number of trainees who completed accredited training	535	280 from webinars, 255 from I-DSO event
4.2c	Number of countries of origin of trainees who completed accredited training	43 countries	43 countries represented in I-DSO symposium, alone.

Endo-ERN ePAG Evaluation Report 2024




We thank our ePAGs for their efforts in 2024!

10 different countries are represented by Endo-ERN ePAGs. Continued efforts need to be made to include input from underrepresented countries in Europe.

- One of the least completed Mandatory activities, inclusion of Endo-ERN logo/ link at their organisation's website, is a focus point for 2025
- Attendance at the Endo-ERN GA is likely difficult for 100% completion as this is a voluntary role and our ePAGs must balance work, health and financial demands associated with travel. Additionally, all MTGs had at least 1 ePAG attending the assembly
- E07 and E09 are more difficult to place due to difficulties in tracking these activities. This requires specific effort in 2025. This holds true for other activities in the Bone category.

Endo-ERN ePAGs per country



Some highlights from 2024:

- Johan de Graaf won 1st prize for his poster on orphan drug shortages at the ERICA RD Conference in Udine, December 2024.
- Arlene Smyth worked tirelessly with TSS UK to create a lay version of **Turners Syndrome CPG**.
- Manuela Brosmle drove efforts to gather input on DSD nomenclature from patients, clinicians and other stakeholders.

Activities	Details	No. of ePAGs completing activity	No. of ePAGs completing all activities
Mandatory	points per event / meeting		



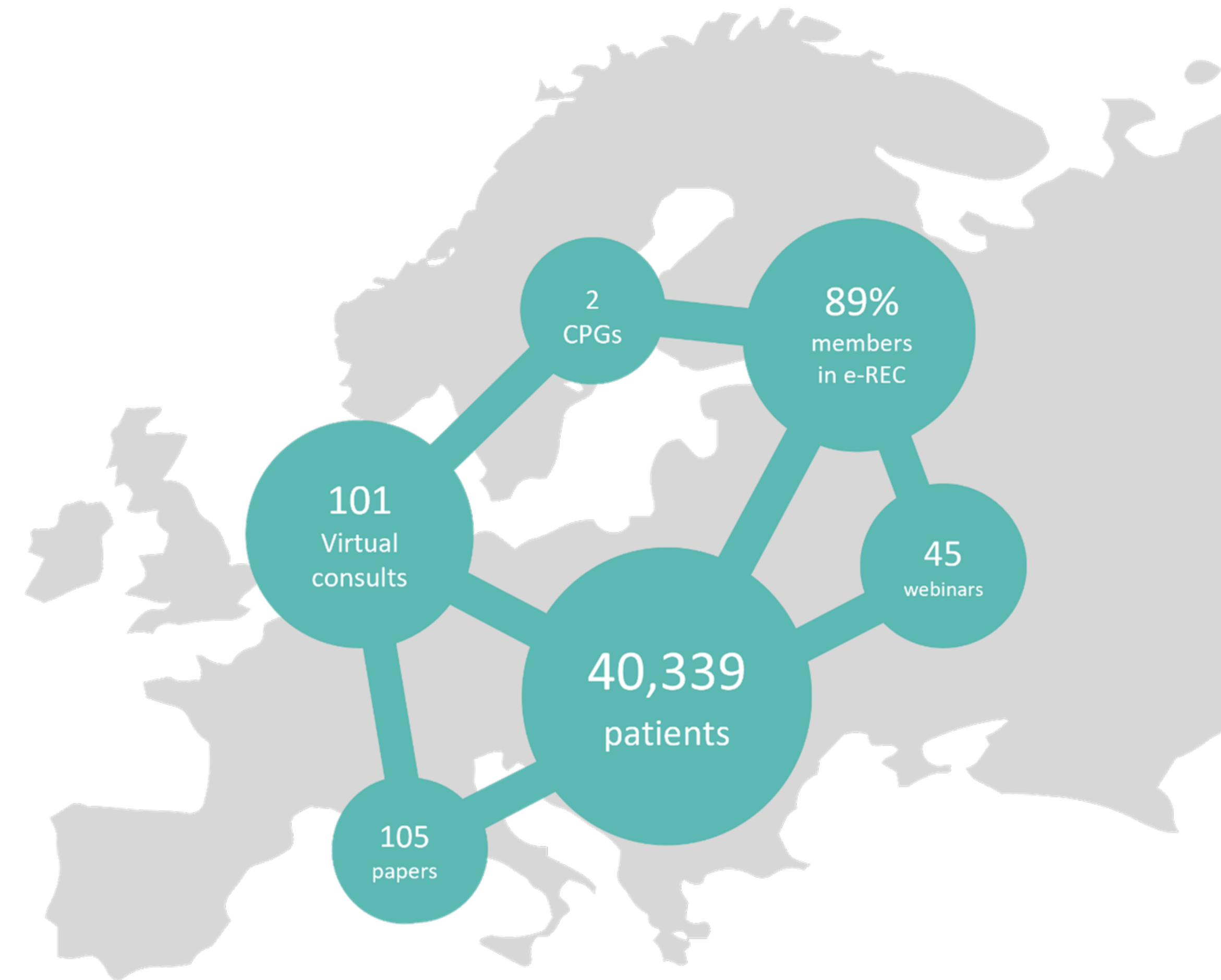
Key reports from 2025

- ERN Monitoring Report 2023/2024
- Endo-ERN Indicators Deliverable (report Y2)*
- Evaluation Report (Y1 &Y2)*
- Endo-ERN ePAG Evaluation Report for 2024 – *ongoing*

Key achievements since ERN Grant onset in 2023

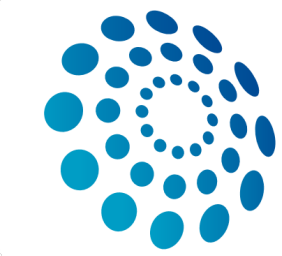


- Established & trained teams in each of our consortium HCPs
- Created and run our HCP performance system
- Obtained accreditation for webinar series
- Promotion of member activities through Endorsement requests
- Migration to CPMS 2.0 system for case consultations
- Built patient & members resources sections of our Website



Endo-ERN member activities since the start of this grant period (October 2023-October 2025).

Areas of Improvement & Goals for 2026



European
Reference
Network



Endo-ERN



WP4 Healthcare & CPMS

Olaf Hiort, Paediatric Chair Endo-ERN

Greta Ginski, CPMS Operational Helpdesk Manager

Emily White, Project Manager & CPMS Operational Helpdesk Manager



Current Statistics



Number of active users in 2025= 254

- 358 user roles

Discussions opened in 2025 = 47

- 302 since ERN started

Number of patients enrolled in 2025 = 72

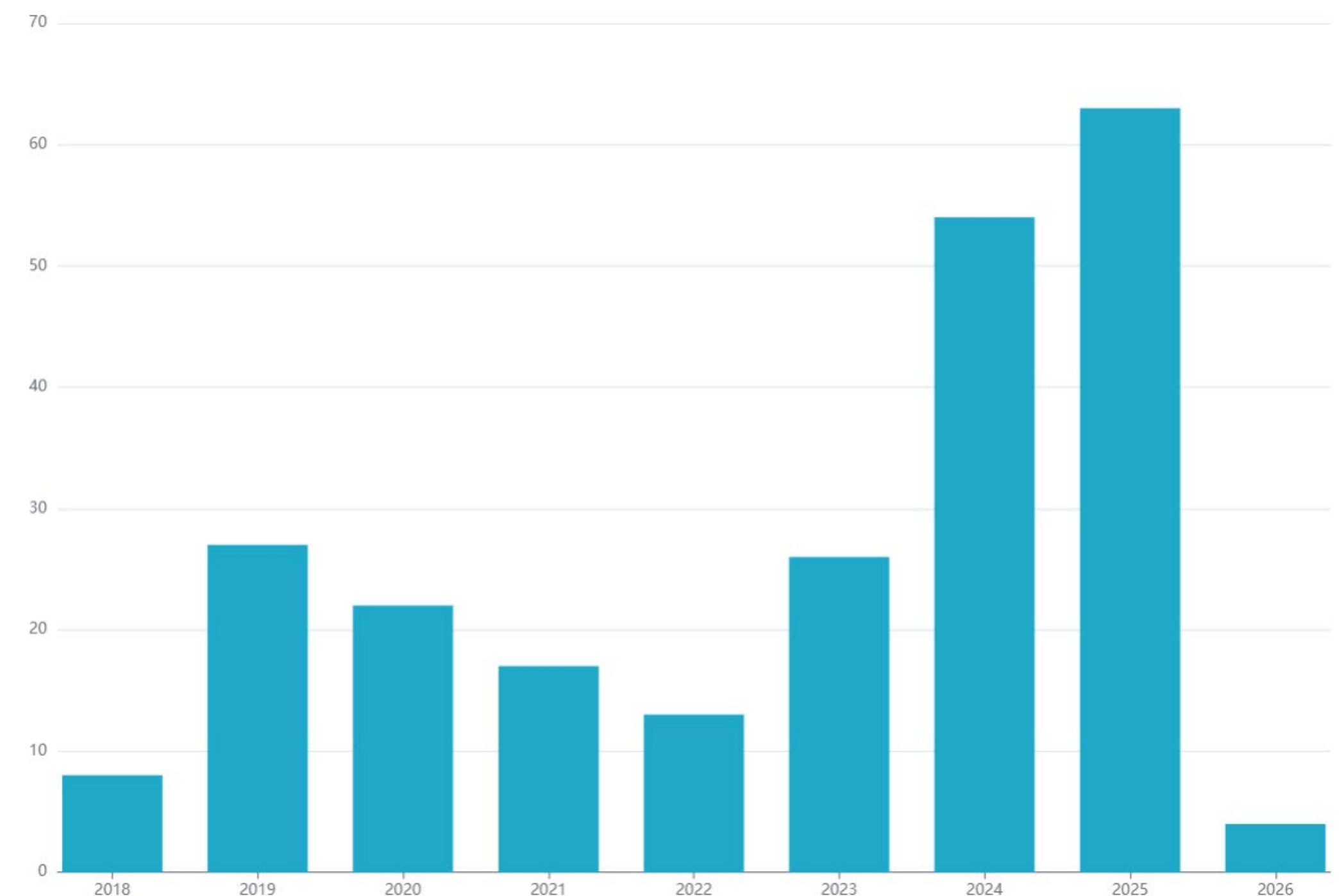
- 967 patients enrolled total

Urgent case discussion = 7

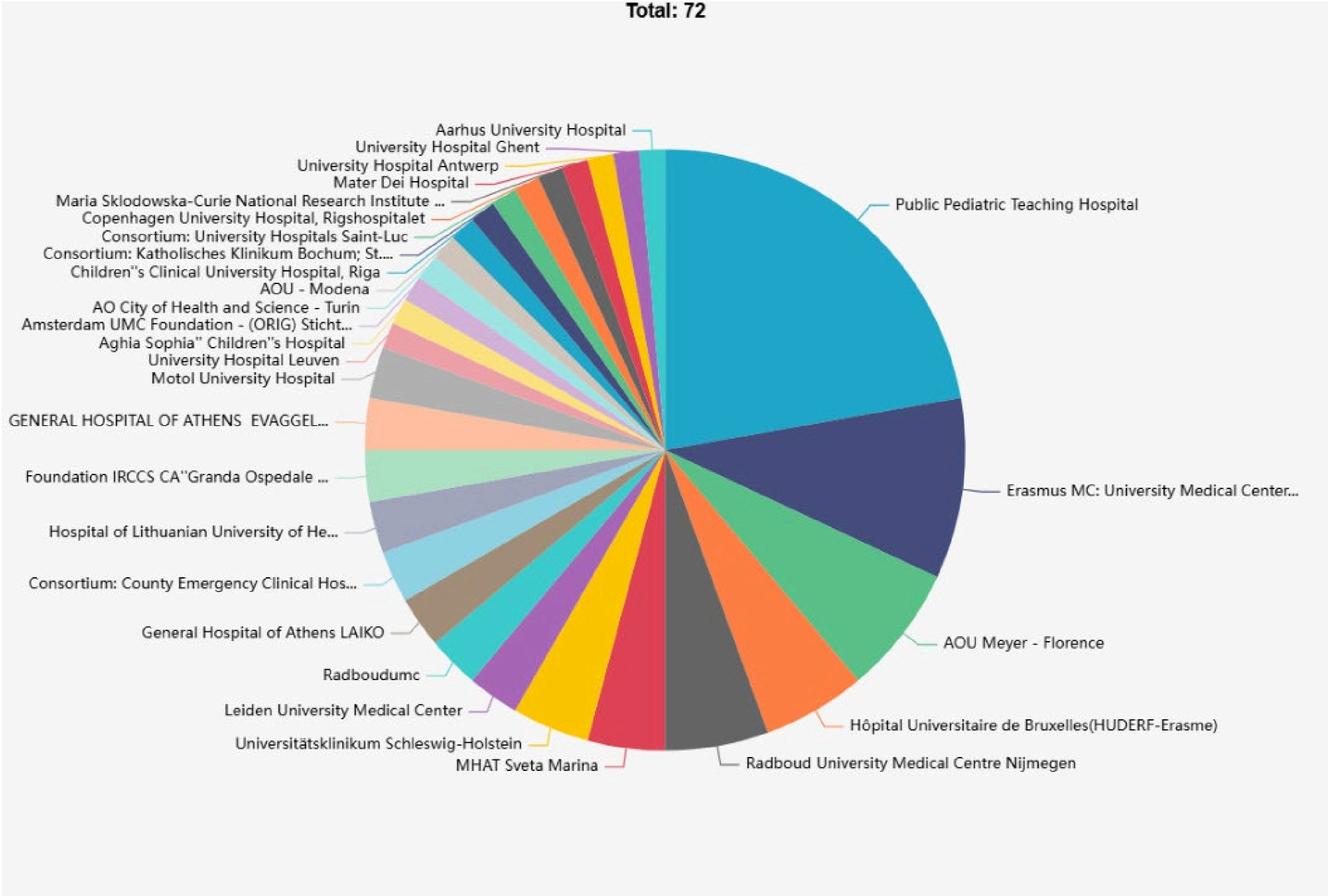
- 55 urgent cases total

Note:
Discussion = 'Panels'

Effective Meetings per year (since start)



Count of patients per HCP in 2025

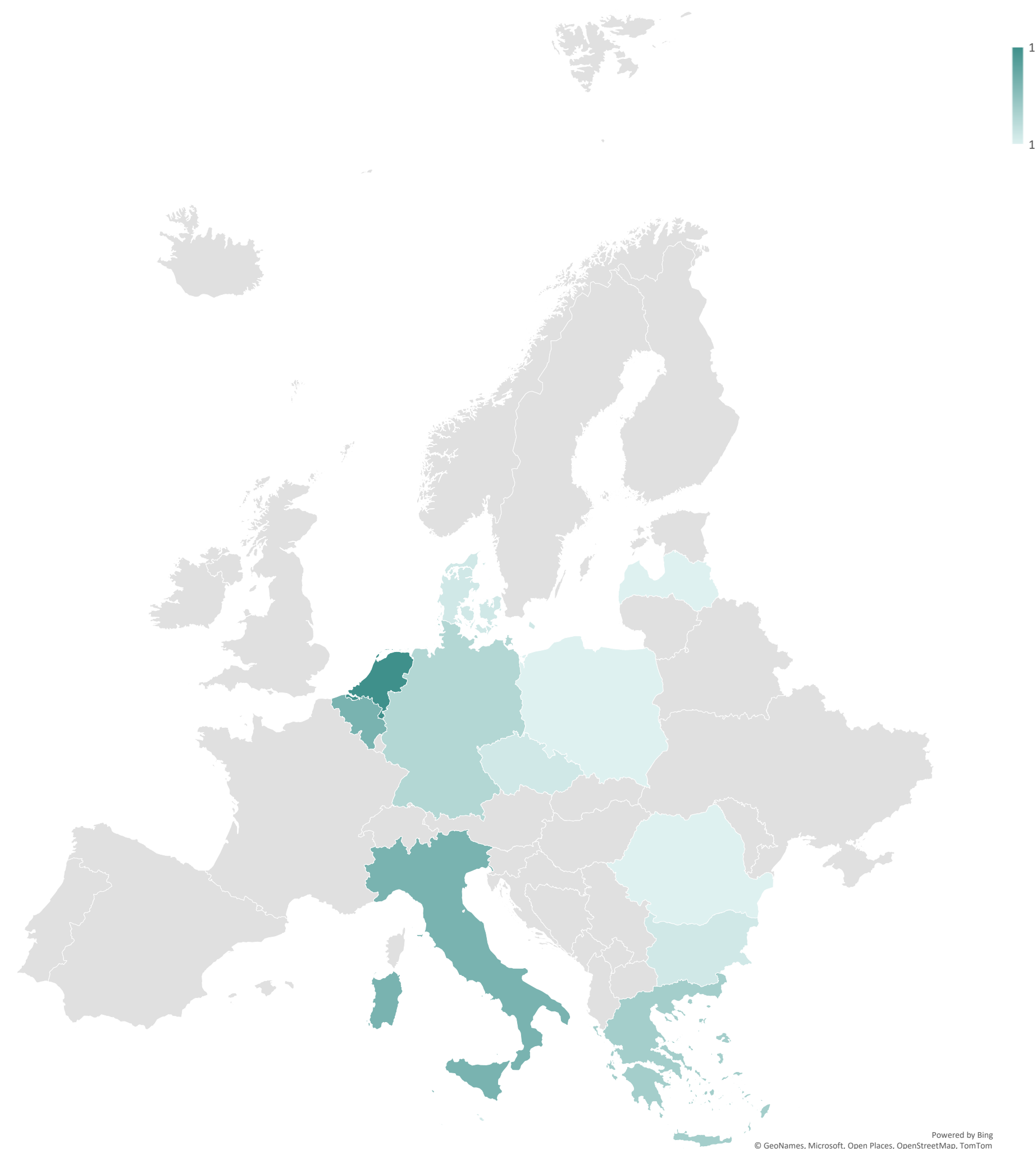


Patient discussions per country 2025

Many HCPs require support with
permission to use CPMS 2.0?

72 patients enrolled
47 discussions opened

Over 70 experts have joined calls
in CPMS and alternative video
calls (Webex) to consult on CPMS
cases






HCPs with permission to use CPMS 2.0 we know of

HCP	Permission obtained
University Medical Centre Ljubljana	DPIA completed
Radboud University Medical Centre Nijmegen	DPIA completed
AOU - Modena	DPIA completed
University Hospital and Faculty of Medicine Tübingen	DPIA completed
Helsinki University Hospital (HUS)	DPIA completed
Universitätsklinikum Schleswig-Holstein	DPIA completed
AOU Meyer - Florence	Case was uploaded, permission assumed
Foundation IRCCS CA'Granda Ospedale Maggiore polyclinic - Milan	Case was uploaded, permission assumed
General Hospital of Athens LAIKO	Case was uploaded, permission assumed
Leiden University Medical Center	DPIA approved, ICT pending
Children's Clinical University Hospital, Riga	Case was uploaded, permission assumed
Erasmus MC: University Medical Center Rotterdam	DPIA completed
Amsterdam University Medical Centers Location AMC	DPIA completed
Spitalul Clinic Judetean de Urgenta Cluj (Cluj County Emergency Clinical Hospital)	Case was uploaded, permission assumed
Hôpital Universitaire de Bruxelles(HUDERF-Erasme)	Case was uploaded, permission assumed
Aarhus University Hospital	DPIA completed
Maria Sklodowska-Curie National Research Institute of Oncology (MSCNRIO)	Case was uploaded, permission assumed
AO City of Health and Science - Turin	Case was uploaded, permission assumed
Katholisches Klinikum Bochum	Case was uploaded, permission assumed
AO City of Health and Science – Turin	Case was uploaded, permission assumed
MHAT "Sveta Marina"	Case was uploaded, permission assumed

Currently updating via
Continuous Monitoring
survey

CPMS 2.0 Goes Mobile

New CPMS 2.0 App Now Available for Download

-  Consult & join meetings on the go
-  Real-time notifications for messages & updates
-  Secure communication with two-factor authentication
- ✓ Respond during rounds or while traveling – no desktop needed

Goal for GA – all attendees download the CPMS 2.0 app & check their EU login app functionality



 Google Play



 App Store





CPMS 2.0 Goes Mobile!

Secure, efficient, and user-friendly case discussions across the



Available in


 Google Play  Apple Store



←↻🏠

https://cpms2.ern-net.eu/screen/meetings

☰

 CPMS 2.0 - CARE

🏠 Home

📁 Patient Record

➕ Add New Patient

👥 Meetings

📄 Supporting doc...

🔍 Advanced search

From

19/01/2026📅

To

Select a date📅

Apply

Reset

51 upcoming meetings

Title ⬆️⬆️	Time	Date ⬆️
Meeting on patient Poppet Jane 830	12:00 - 12:30 30 min	22 Jan 2026
🔄 MTG3 CPMS 2.0 Meeting Endo-ERN	13:30 - 14:30 1 hr	28 Jan 2026
🔄 MTG3 CPMS 2.0 Meeting Endo-ERN	13:30 - 14:30 1 hr	28 Jan 2026
🔄 MTG1 CPMS 2.0 Meeting Endo-ERN	15:00 - 16:00 1 hr	03 Feb 2026
🔄 MTG7 CPMS 2.0 Meeting Endo-ERN	16:00 - 17:00 1 hr	04 Feb 2026
🔄 MTG6 CPMS 2.0 Meeting Endo-ERN	13:00 - 14:00 1 hr	20 Feb 2026
🔄 MTG3 CPMS 2.0 Meeting Endo-ERN	13:30 - 14:30 1 hr	25 Feb 2026
🔄 regular DSD case-conference	16:00 - 17:00 1 hr	03 Mar 2026
🔄 MTG7 CPMS 2.0 Meeting Endo-ERN	16:00 - 17:00 1 hr	04 Mar 2026
🔄 MTG2 - CPMS meeting Endo-ERN	14:00 - 15:00 1 hr	11 Mar 2026
🔄 MTG6 CPMS 2.0 Meeting Endo-ERN	13:00 - 14:00 1 hr	20 Mar 2026

CPMS 2.0 - 1.72.3



Recurring MTG meeting in CPMS - status

[CPMS Recurring Meetings - Endo-ERN](#)

No MTG4 or MTG5 – please coordinate with heldesk for preferred meeting times

New Support Feature



1/1 Support

Running into issues or have questions on CPMS 2.0?

--> Use the [booking software](#) on our website to schedule a 15-min call with the Operational Helpdesk

- Index
- Virtual Consultations
- CPMS 2.0
- Log-in
- HCP Permission&Consent
- Manuals&Guides
- CPMS Recurring Meetings
- Ukraine
- | 1/1 Support

Schedule a Meeting

CPMS 2.0 helpdesk Endo-ERN

CPMS 2.0 Support Call ▶

POWERED BY
Calendly



Work Package 5

EuRREB Registries, Data Management and Analysis

Update on activities

Natasha Appelman-Dijkstra

EuRREB Coordinator



Funded by
the European Union

Endo-ERN members activity

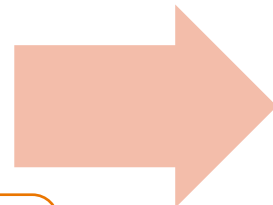


e-REC active centres

Grant indicator:
75% by 2027

December 2024

- 87 centres (82%)



December 2025

- 97 centres (92%)

New cases in 2025*

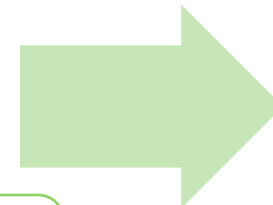
13185

Core Registry centres with cases

Grant indicator:
50% by 2027

December 2024

- 31 centres (29%)



December 2025

- 44 centres (42%)

1049



e-REC changes 2026

1) MTG6

↳ [Pituitary adenoma](#) ORPHA:99408 -

↳ [Functioning pituitary adenoma](#) ORPHA:314753 -

↳ [Cushing disease](#) ORPHA:96253

↳ [Functioning gonadotropic adenoma](#) ORPHA:91348

↳ [Mixed functioning pituitary adenoma](#) ORPHA:314759 +

↳ [Nelson syndrome](#) ORPHA:199244

↳ [Prolactinoma](#) ORPHA:2965

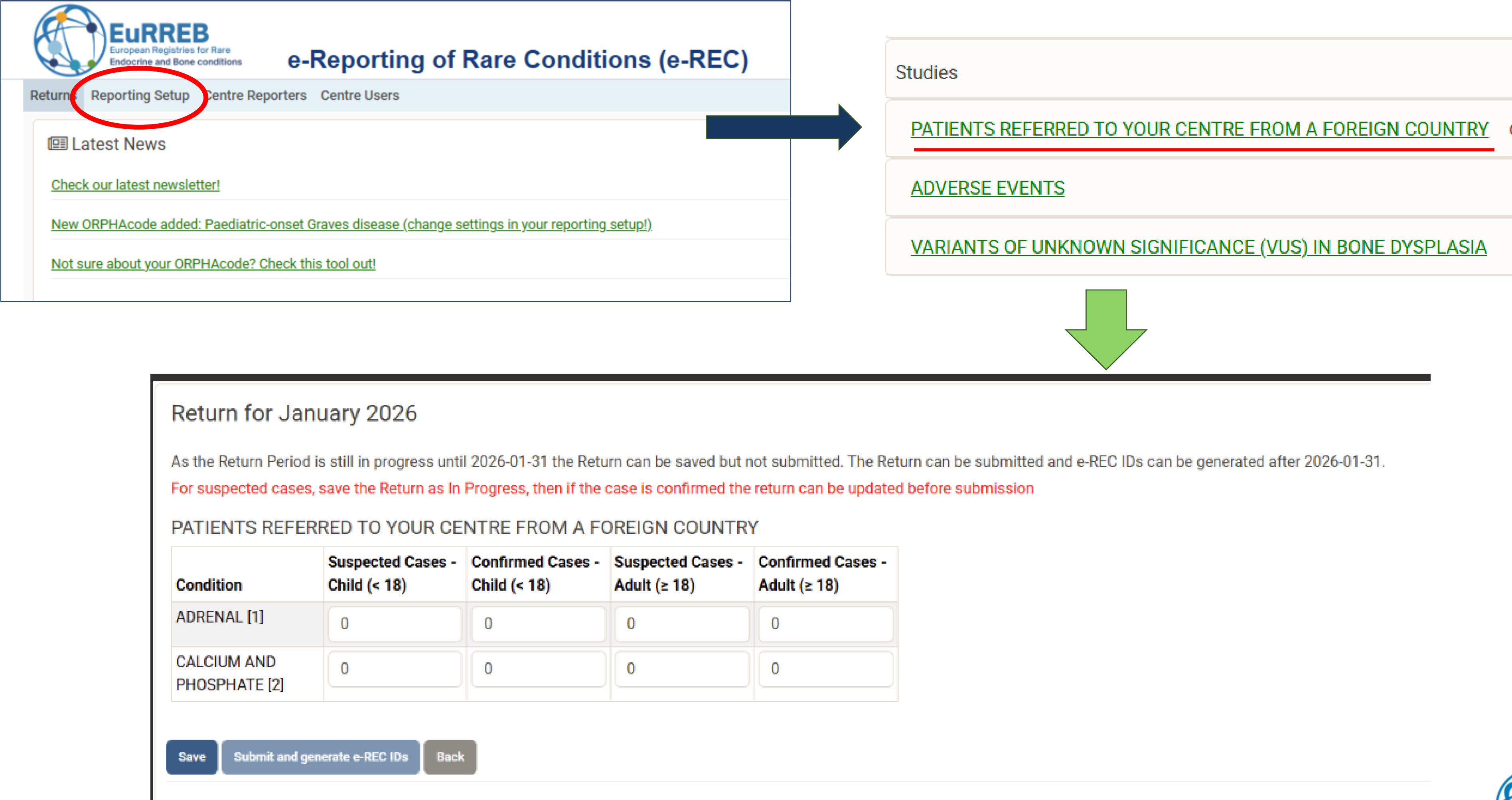
↳ [Somatotropic adenoma](#) ORPHA:96256 +

↳ [TSH-secreting pituitary adenoma](#) ORPHA:91347

} Other types
of pituitary
adenomas



e-REC changes 2026



The CR Platform updates



- 1) Autosave functionality (CSMs)
- 2) Direct links from the condition-specific modules to generic PROMs

- 3) New category – Life-course outcomes

Diagnosis-specific Outcome Details

Patient ID - 427 Date of Birth - 1959-10-27 Condition Group - Bone Dysplasia

[Supplemental material to complete this module](#)

Questionnaire: Fibrous dysplasia/McCune Albright module - V4_Apr_2024

Created Date: 2024-06-21

Completed By: Sophie Huisman

Linked Generic Outcomes

WHO-ICF Mobility

MSK-HQ

BPI-SF

NPSI



Core Registry

Dashboard Patients Centres Users Centre Users Patient Users Reference Data Emails Reporting Mariya Cherenko - Leiden University Medical Center

Life-course Outcomes Patient ID - 4291 Date of Birth - 2011-11-11

* - Condition sections marked with * contain mandatory fields that require your attention!

Core Data * BONE DYSPLASIA Generic Outcomes Diagnosis-specific Outcomes Life-course Outcomes Documents Audit Patient List

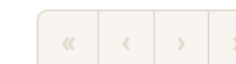
Pregnancy

10 entries per page

Search:

Created Date Latest Assessment Date Completed By Questionnaire Actions

No Outcomes found



New PROMs

- 1) Cancer worry scale (MEN-1 oriented)
- 2) HPQ-28 (Hypoparathyroidism)
- 3) HPES-Impact and HPES-symptoms (Hypoparathyroidism) - WIP



Other activities:

- With WP7: KPIs from guidelines
- Genetic testing in hypoparathyroidism (cross-country project)
- Case study, EuRREB/EC Discussion Paper on ERN Registries
- January EuRREB Newsletter



WP6 Education and Training

Violeta Iotova, Kami Markovska, Sevim Zekerries,
Manuela Brösamle, Petra Bruegmann



Endorsements

- 4 endorsed events for 2026 so far
- **NOTE** for those interested in receiving an Endorsement:
 - General requirements of the Network
 - Follow-up info required

Members can apply for endorsement for their locally organised educational events

Endorsed events are promoted on the Endo-ERN channels

Application should be received at least 6 weeks before the event

All criteria can be found on the website: <https://endo-ern.eu/members/endorsement/>

Clinical Exchange

- 8 exchanges/1 week were available for 2026
- 4 approved exchanges- 6 weeks
- First Exchange is in April
- Longer exchange periods- 2 participants are going for 2 weeks

Webinars& Accreditation



Webinars

- **REMINDER:** MTGs need to provide a minimum of 2 webinars per year-omitting summer months (July-August)
- Organizing a webinar needs to be reported to WP6 at least 6 weeks in advance
- **NOTE:** A webinar will be processed after the Webinar Planning Form is submitted

Organized Webinars

- **MTG5-** An update on genetics and clinical management of tall stature
- **MTG6-** Corticotroph tumors and Postoperative Quality of Life – Perspectives from a Neurosurgeon, an Endocrinologist and a Patient
- **Joint EYE/Endo - Genetic basis of BPES** (*Blepharophimosis, Ptosis, and Epicanthus Inversus Syndrome*) and implications for genetic counseling

Accreditation

- Accreditation means more registrants and attendees
- Minimum 5 webinars per accreditation pack

←

→

↺

🏠

https://endo-ern.eu/webinars/

🔊

☆


⚙️

👤

⋮

🖥️

European
Reference
Network



Endo-ERN

Rare Endocrine Conditions

About Us

For Professionals

For Members

For Patients

🔍

🏠 / Webinars

Index

Upcoming events

Webinars

Registration

Previous events

Overview of Webinars



Showing 106 entries

🔍

MTG	Theme / Title	Date	Time CET	Speaker(s)	Register	View	CPMS
	ESE Talks Webinar...Recent therapeutic perspectives for rare endocrine diseases (multiple examples)	2025-11-04	17:00 – 18:30 hr		👤+		
MTG2	Webinar on the Clinical Practice recommendations for the diagnosis and management of XLH	2025-10-29	17:00 – 18:00 hr		👤+		
MTG5	An update on primordial dwarfism conditions webinar	2025-10-20	15:00 – 16:00 hr		👤+		
MTG8	Diagnosis and management of TSH resistance	2025-10-07	17:00 – 18:00 hr	Edward Visser and Luca Persani	👤+		
MTG4	Genotype-phenotype correlations in CAH	2025-09-24	16:00 – 17:00 hr		👤+		
MTG7	Advances in the diagnosis and management of Congenital Hypogonadotropic Hypogonadism (CHH) webinar	2025-09-22	17:00 – 18:15 hr		👤+		
	ESE Talks Webinar...CAH: diagnostic and therapeutic challenges across the lifespan	2025-09-02	17:00 – 18:30 hr		👤+		
	Rare disease research and the value of registries	2025-06-27	16:00 – 17:00 hr		🔒	👁️	
MTG6	Congenital combined pituitary hormone deficiency in Europe webinar	2025-06-25	16:00 – 17:00 hr		🔒	👁️	
MTG6	Cushing's Syndrome Webinar	2025-06-10	16:00 – 18:00 hr	Jérôme Bertherat, Marta Baranska and	🔒	👁️	

RED Winter School

- Program built on MTG3; MTG5; MTG6 and MTG7
- We are expecting 36 highly qualified students in Varna for the Winter School dedicated to Rare Endocrine Diseases (RED)
- 30/36 are from ERN
- 20/36 are from The Netherlands, Italy, Ireland, France, Czech Republic, Latvia, Lithuania, Belgium, Turkey
- 16/36 are Bulgarians
- 24 distinguished teachers



**RARE ENDOCRINE DISEASE
WINTER SCHOOL VARNA**

RED Winter School

28.02 - 3.03.2026

Sts. Constantine and Helena Resort, Varna, Bulgaria
Ensana Aquahouse Hotel

Financed by the European Union-NextGenerationEU, through the National Recovery and Resilience Plan of the Republic of Bulgaria, project № BG-RRP-2.004-0009 "Medical University – Varna: Enhancement of Translational Excellence Achievement in Medicine (MUVE-TEAM)"

WP7 Clinical Practice Guidelines & Clinical Decision Support Tools

Olaf Dekkers, Anna Nordenström, Charlotte Gaasterland, Alessandro Lazzerini,
Diana Vitali



Funded by
the European Union

In memoriam: Jette Kristensen



Points to discuss

- 3 ongoing CPG's
- Endorsement strategy

3 ongoing CPGs

- **Congenital hypopituitarism:** First working group meeting in February
 - Survey to find out the most controversial topics
- **Hypoadrenalism replacement therapy:** First meeting with chairs in January
- **Hypothalamic obesity:** Chairs are assigned

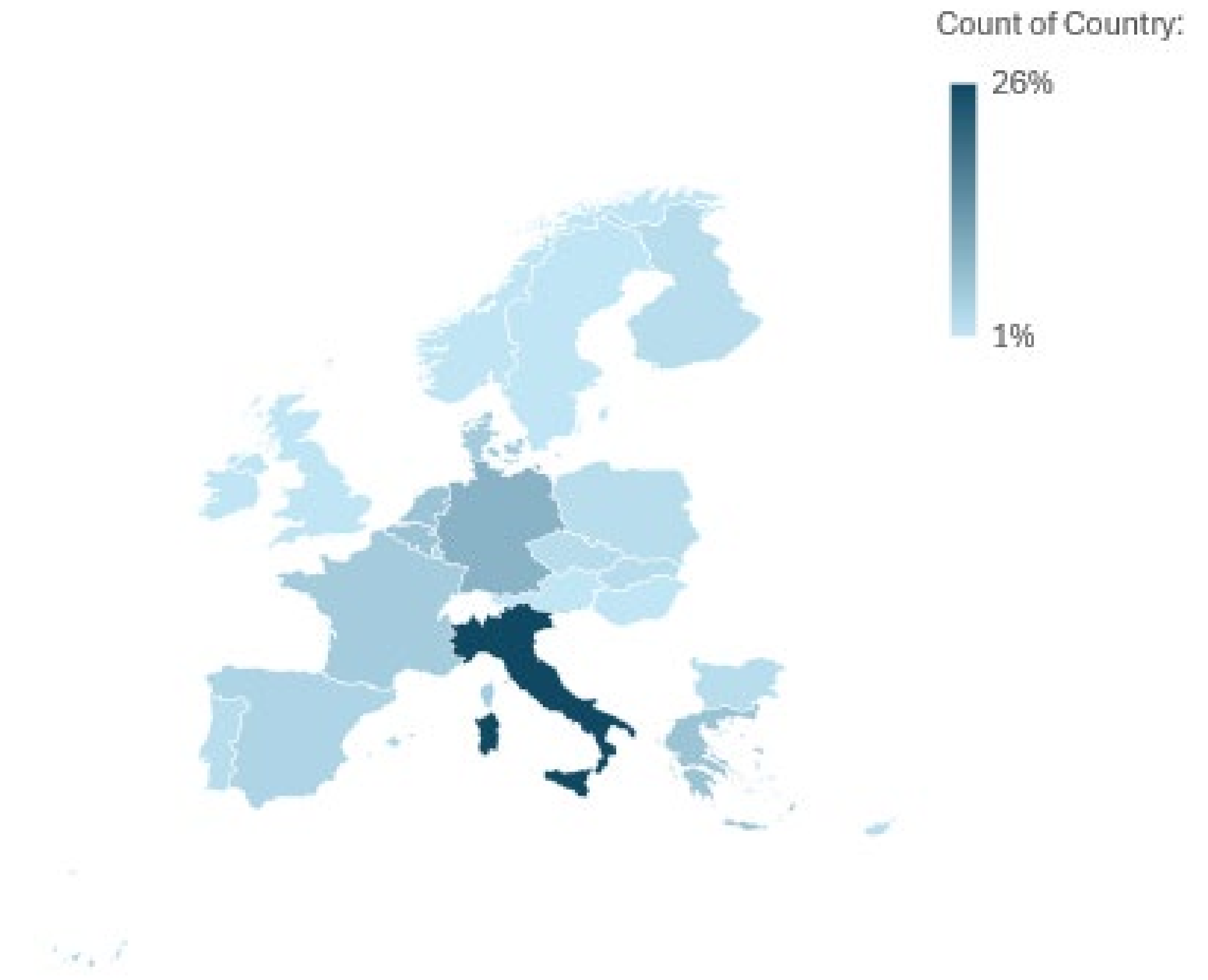
Endorsement strategy

- Current strategy drafted by Charlotte G
- Survey for input sent out early May, deadline 20 June 2025
- Updated strategy to be discussed in Athens



Submissions – 77 total

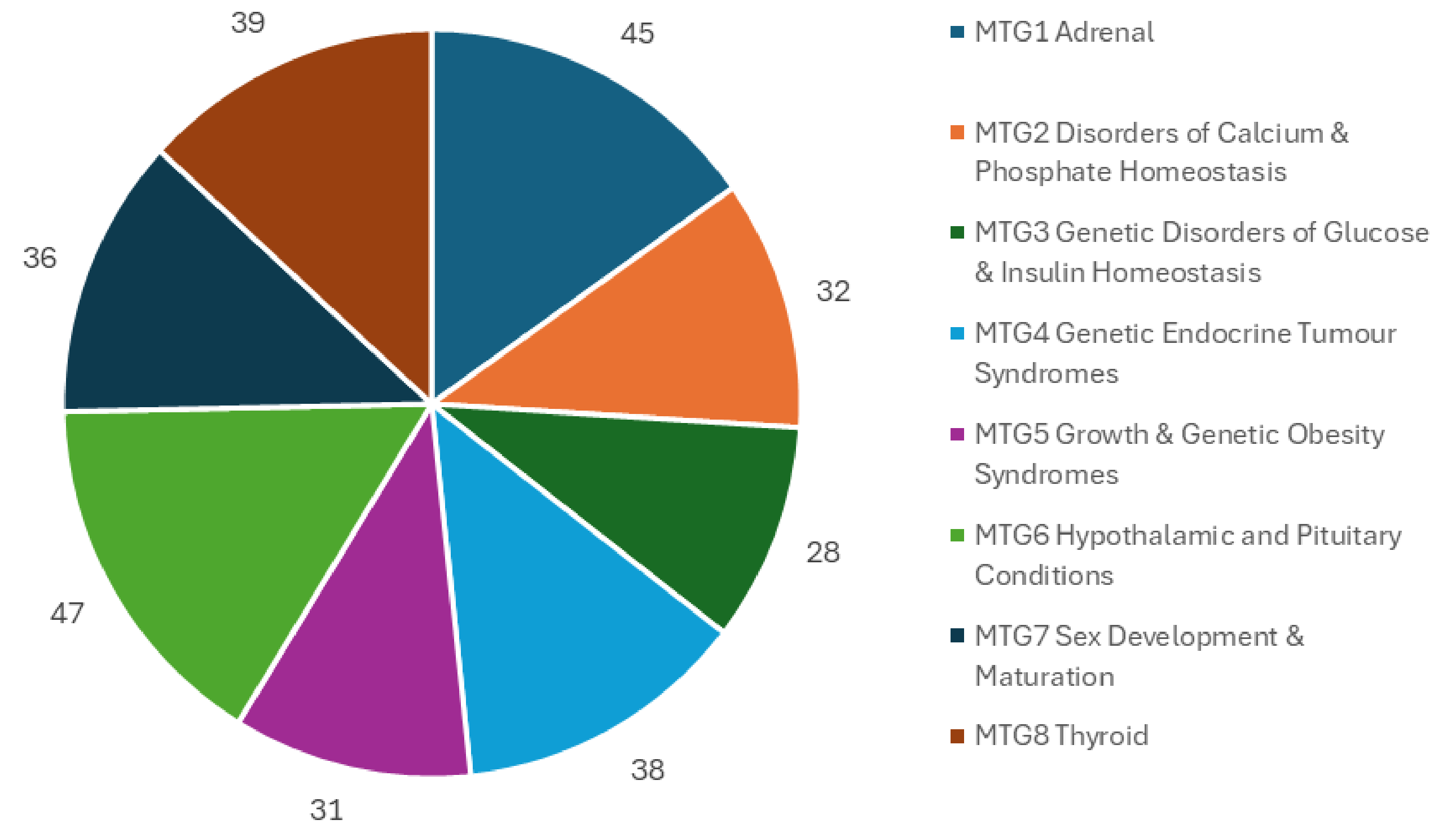
Austria	1%
Belgium	6%
Bulgaria	3%
Cyprus	3%
Czech Republic	3%
Denmark	5%
Finland	3%
France	5%
Germany	9%
Greece	6%
Hungary	1%
Republic of Ireland	1%
Italy	26%
Luxembourg	4%
Malta, Montana	1%
Netherlands	6%
Norway	1%
Poland	3%
Portugal	3%
Slovakia	3%
Spain	4%
Sweden	1%
United Kingdom	1%



Results demographics

- Majority main or sub representatives
- 1 patient association (from Italy)

MTG representation





What do you think endorsement of a document by Endo-ERN should mean?



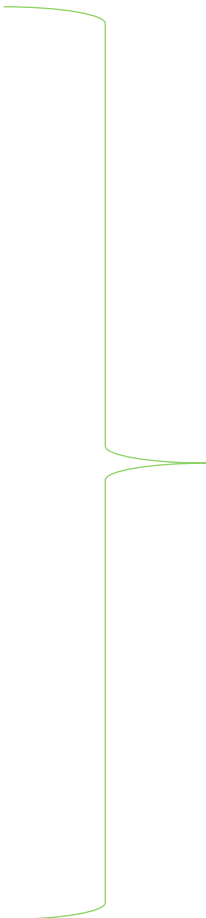
The document's recommendations should be followed by Endo-ERN members	29%
The document's recommendations should be known by Endo-ERN members	51%
The Endo-ERN members should be aware of the document	20%

*My understanding of endorsement of a document by EndoERN should mean, is that the document/meeting should be **supported or approved by EndoERN**. Therefore EndoERN should know about the document's **recommendations** and approve its publication.*

that the document has been reviewed by recognized experts within the network and reflects a consensus or broad agreement among specialists in the field.

Endorsement should be given after consultation and possible comments/feedback from Endo-ern members

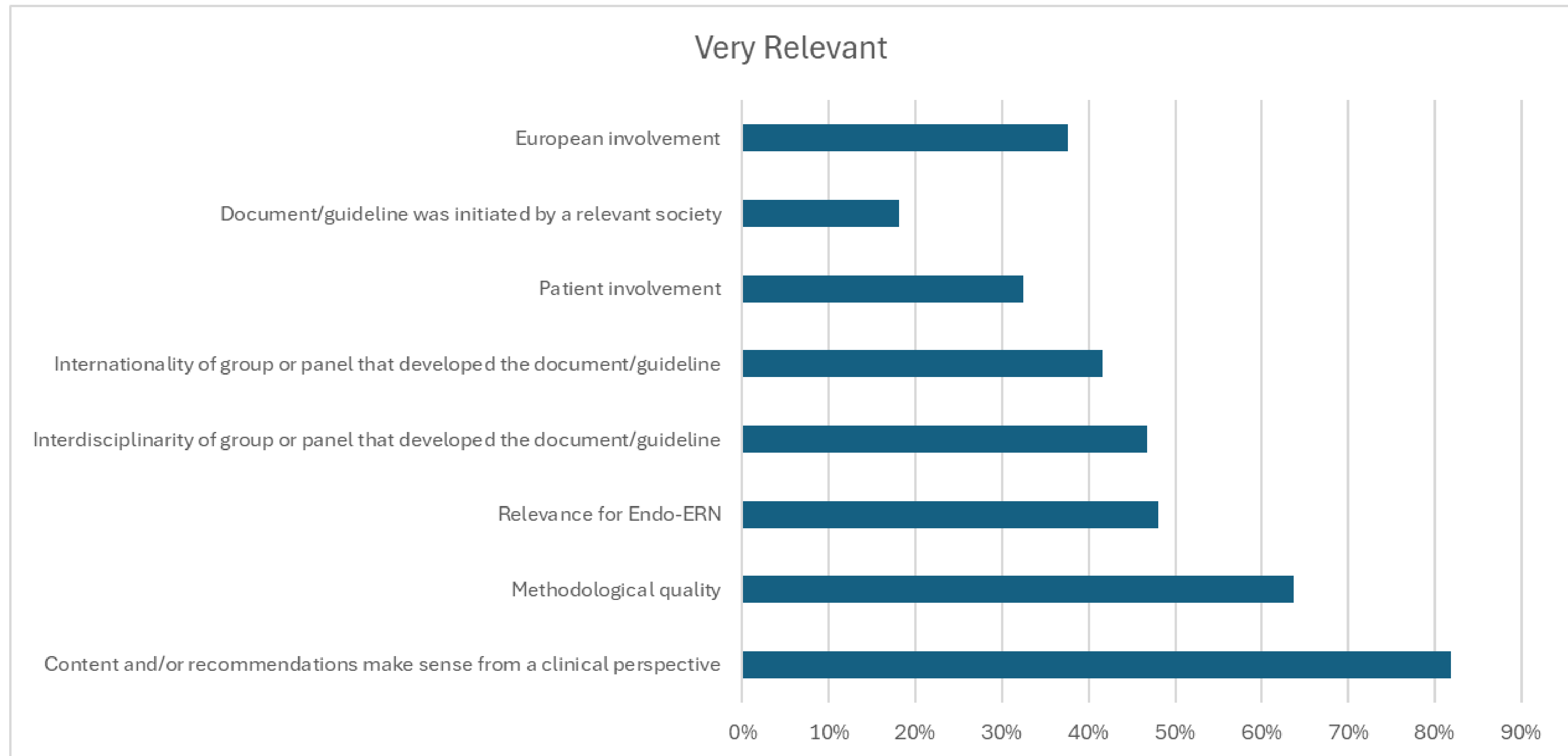
Endorsement means that EndoErn recommend it because 1) the methodology is correct or 2) the topic covered is consistent with the EndoERN mission



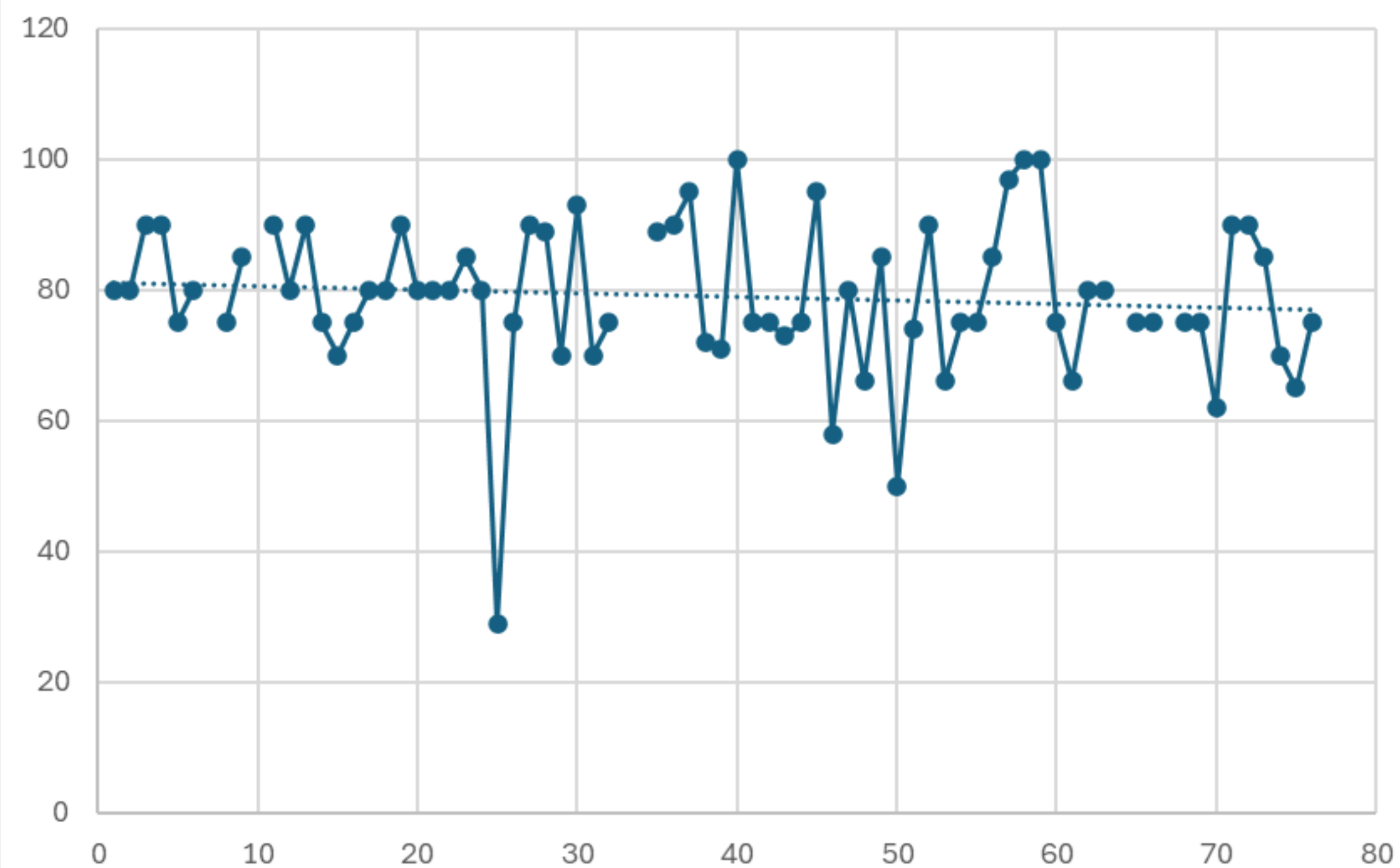
"ENDO-ERN endorsement enhances the document's importance. It means that the ENDO-ERN supports such a document. (support/acceptance AND compliance/adherence)"



Please rate how relevant you find these criteria for the endorsement of consensus documents/guidelines:



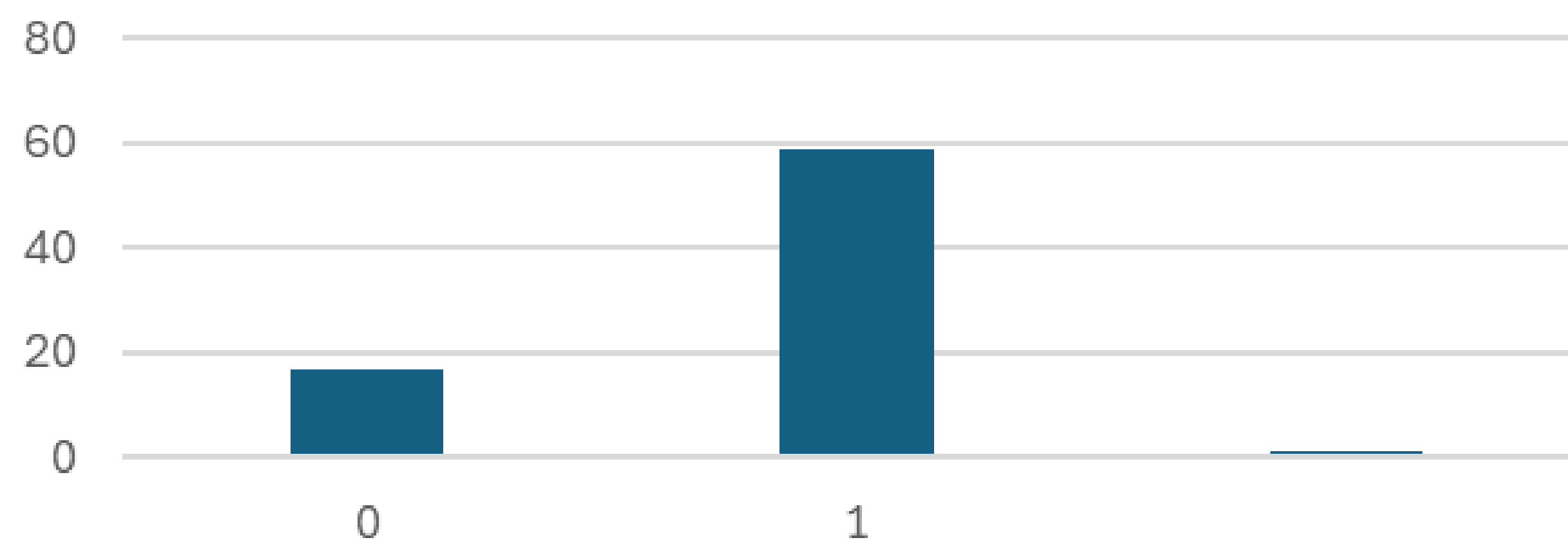
When a document provides recommendations, how much/many of the recommendations would you have to agree with from a clinical point of view before you would consider the document good enough for endorsement by Endo-ERN?



Do you think that the
endorsement strategy for clinical practice
guidelines should be made in alignment
with all ERNs?

Yes = 1

No = 0



Delphi on minimum set of quality criteria

- Representation from many ERN's
- Consensus meeting only yesterday
- CPGs versus consensus statements



WP8/9 Capacity building, best practice sharing & other activities for Ukraine

Charlotte van Beuzekom, Project Manager Endo-ERN



Funded by
the European Union

Joint-ERN Information Webinar



Improving Rare Disease Care in
Ukraine: Access to ERNs and
Expert Resources

presented by

Endo-ERN & ERN eUROGEN

Wednesday 18 February 2026 16:00-17:45 CET

- Dedicated page for Ukrainian clinicians – and we are developing a database of contacts
- Upcoming joint webinar with eUROGEN
- Clinical Exchange has a Ukrainian participant in 2026
- 2026 AI translations of the existing webinar library and potentially other assets



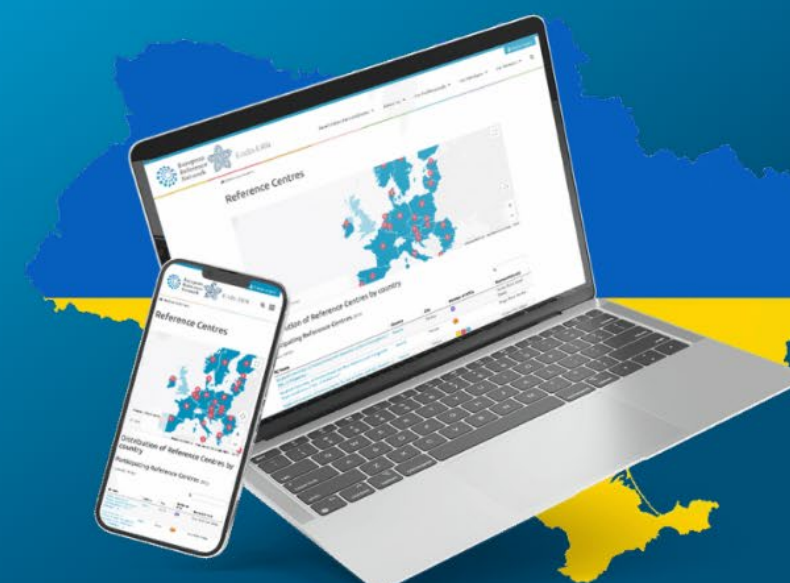
Розкажіть нам про себе

Підтримка українських лікарів за допомогою Endo-ERN

Endo-ERN прагне допомогти українським лікарям зв'язатися з провідними європейськими експертами з рідкісних ендокринних захворювань. За допомогою Endo-ERN ви можете запросити віртуальні консультації з експертними центрами по всій Європі, отримати доступ до клінічних рекомендацій, заснованих на спільному європейському досвіді, та взяти участь в освітніх та тренінгових програмах, розроблених для клініцистів.

Ваш досвід як лікуючого лікаря залишається ключовим – Endo-ERN лише сприяє тому, щоб європейські знання та підтримка безпосередньо допомагали вашим пацієнтам. Процес відбувається за допомогою спеціального хабу, розташованому у лікарні «ОХМАТДИТ» у Києві.

Знайти експерта



Transversal Working Group on Genomic Testing

Thomas Eggermann (University Hospital Aachen, Germany)

Dirk Prawitt (Center for Paediatrics and Adolescent Medicine, University Medical Center, Mainz, Germany)



Funded by
the European Union

Status

Currently 31 participants from 10 different countries

Virtual meetings: every 2-3 months, with the current focus on the joint paper

ESHG – ERN collaboration

- communication managers (E. White) and liaison managers (TE, DP)
- Virtual meetings every 3 months

Teaching & Masterclass

- ESHG becomes part of the ERNs course portfolios and vice versa
- Germline genetics needs to be part of the educational program of ERNs
- ESHG is willing to engage in education if the part of genetics is part of the ERNs educational programmes
- ERN may request collaboration from ESHG to support genetics-related educational contents (“not related to Industrial interests”).
- Developing ESHG-ERN Courses endorsed by ESHG and ERN.

Cross-referencing in ESHG and ERNs websites regarding collaborative actions, joint-educational initiatives and liaison members.

Visibility & Communication

- **Visibility : ERN visible on the ESHG website and vice versa**

How to make ERNs visible on the ESHG website?

How to make ESHG visible on the ERN Websites?

Visibility of ERN-ESHG liaison members on both websites?

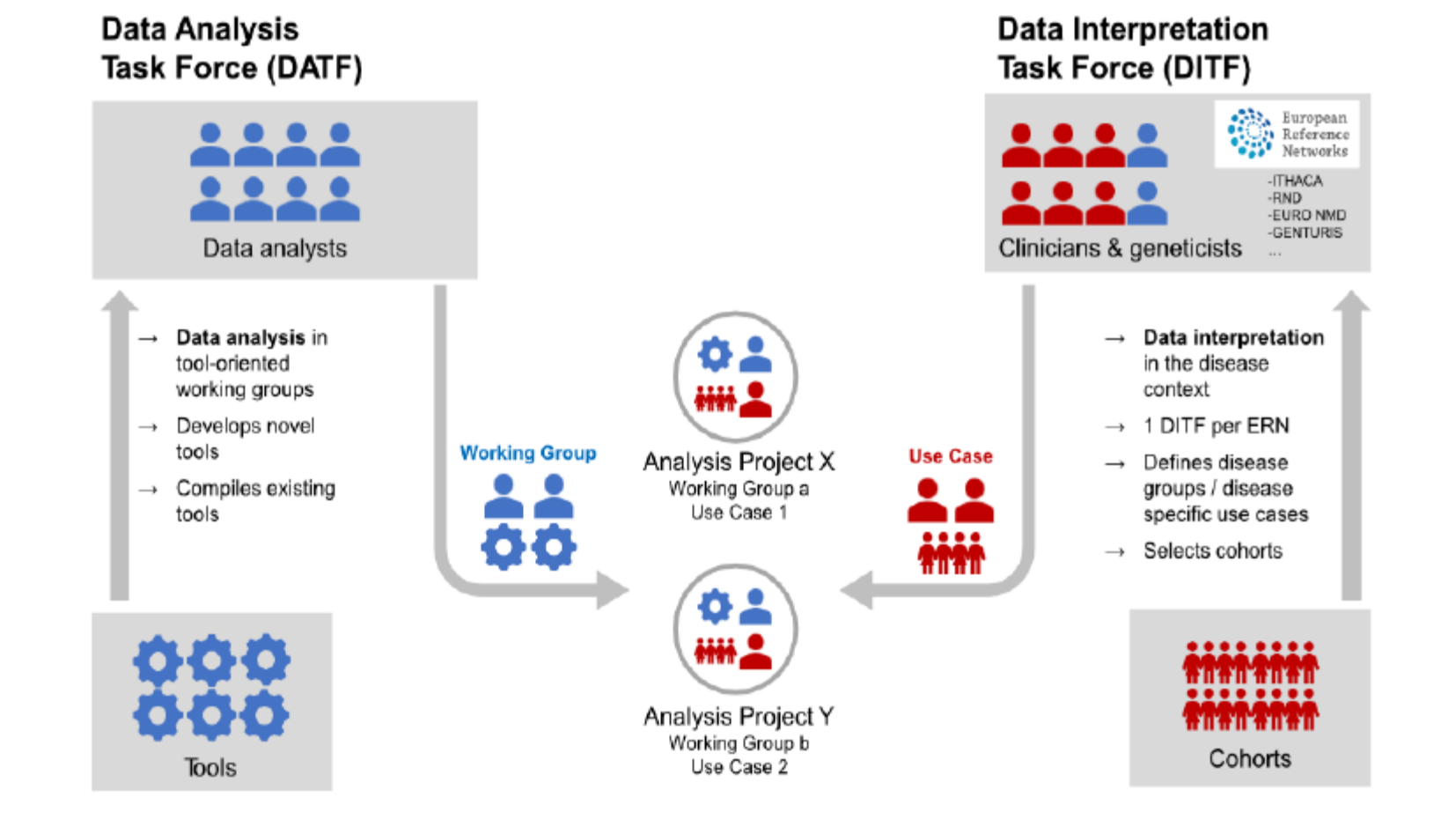
Communication managers (for ERN-ESHG information flux) and inform the ESHG Office (Jerome del Pichia) ?

Update ERDERA Diagnostic Research Workstream

Joint expert re-analysis

WHAT CAN YOU DO AS ERN TO CONTRIBUTE TO THE DRW?

- Inform your HCPs to contribute samples to the DRW
- Have HCPs sign up to contribute to the ERN-specific DITF if they have expert knowledge on:
 - ✓ Data interpretation for (non-)coding variants
 - ✓ Specific genes or phenotypes of relevance to your ERN
 - ✓ Novel gene/mechanism/paradigm discovery
 - ✓ (Data analysis to join DATF working groups)
- Nominate a candidate for the position ERN-specific DITF lead and **provide the information to Holm Graessner and Lisenka Vissers**



The WG Genetic Testing aims to moderate the cooperation between the HCPs of our network and the ERDERA Diagnostic workstream group, e.g. by bringing them in contact with ERDERA, and by exchanging information and experiences on national level in respect to the legal issues (e.g. data protection).

In addition to these individual cooperations between our HCPs and ERDERA Diagnostic Workstream, our ENDO-ERN has to assign colleagues for the Data Interpretation Task Force (DIFT).

We have been advised to encourage the MTG chairs to promote this in the network. If all agree, the follow up dissemination to HCPs and identification of Endo-ERN representation for the Data Interpretation Task Force, can be done afterwards by our coordination office.

Is it possible to ask each MTG to nominate at least one representative to discuss further steps with the WG (deadline: Feb. 15th 2026).

ERDERA2026 call

Conceptual phase of Calls for Proposals 2026

“Resolving unsolved cases in rare genetic and non-genetic diseases”

Submission deadline for pre-proposals: February 12th, 2026 at 2 PM (CET)

Currently pre-proposal discussions on

CHH (Marco Bonomi)

RTH (Luca Persani)

TWG on genomic testing tries to support these

(DSD (Martin Kircher))

Joint paper (in preparation): „What the endocrinologist expects from genetic testing and reporting“



Contents:

	Topic
1	When is a genetic test appropriate, including regulatory aspects, when is it allowed?
2	Standardised genetic testing procedures (cytogenetics, molecular cytogenetics/array, single gene analysis vs targeted panel vs WES vs WGS) What is optimal?
3	How to achieve a standardized genetic testing quality?
4	Standardized reports addressing the needs of the clinicians
5	Germline and somatic genetic testing have to be considered
6	How to deal with VUSs, Recall of VUS
7	Incidental/Secondary findings
8	Common research activities
9	How is genetic testing reimbursed across Europe

- First versions of sections 1-3, 5 finished; final versions of sections 4, 6, 7; sections 8 and 9 in preparation.
- Deadline: April 2026, submission in discussion

Status



In preparation:
Matrix overview about the genetic expertise available in the ENDO-ERN: a survey is in preparation

HCP	Country	City	Representative(s)	Accreditation	methods	disease 1	disease 2	disease 3	disease 4	disease 5	relevant papers	Involvement in Guidelines	Testing for research possible
Department of	Cyprus	Nicosia	Leonidas Phylactou, Vassos Neocleous	ISO 15189	NGS, Sanger,	CAH	MEN2	MODY	Hypogonadotropic Hypogonadism		38637882; 31505704; 29396	Yes	Yes
University Hospital	Germany	Aachen	Thomas Eggermann, Miriam Elbracht	ISO 15189	(MS)MLPA, MiSRS	SRS	BWS	upd(20)mat	PWS	TNDM		PMID: 275859	yes
University Medical	Germany	Mainz	Dirk Prawitt, Joachim Pohlenz	ISO 15189	RT-PCR (rearr	Heritable thyr	Endocrine tumor	Abnormal growth, Imprinting Defects (BWS)			32462478, 35	PMID: 332720	yes

Feel free to contact us at any time!

Ideas/Suggestions/Contacts are welcome,
preferably from every MTG

Contact:

Thomas Eggermann
(teggermann@ukaachen.de)

Dirk Prawitt
(dprawitt@uni-mainz.de)

Transversal Working Group on Transition in Care

Andrea Isidori (Azienda Ospedaliero-Universitaria Policlinico Umberto I, Rome, Italy)

Ulla Doehnert (Universitätsklinikum Schleswig-Holstein, Luebeck, Germany)



Funded by
the European Union

Checklists for transition of patients with specific conditions

Final / in review:

MTG 1: Congenital Adrenal Hyperplasia

MTG 2: X-linked Hypophosphatemia

MTG 4: MEN 1

MTG 7: Congenital Hypogonadotropic Hypogonadism

MTG 8: Congenital Hypothyroidism

In preparation:

MTG 5: Prader-Willi Syndrome

MTG 6: AVP Deficiency

MTG 7: Turner Syndrome

8th TALENT Conference meeting



European
Reference
Network



Endo-ERN



SAPIENZA
UNIVERSITÀ DI ROMA

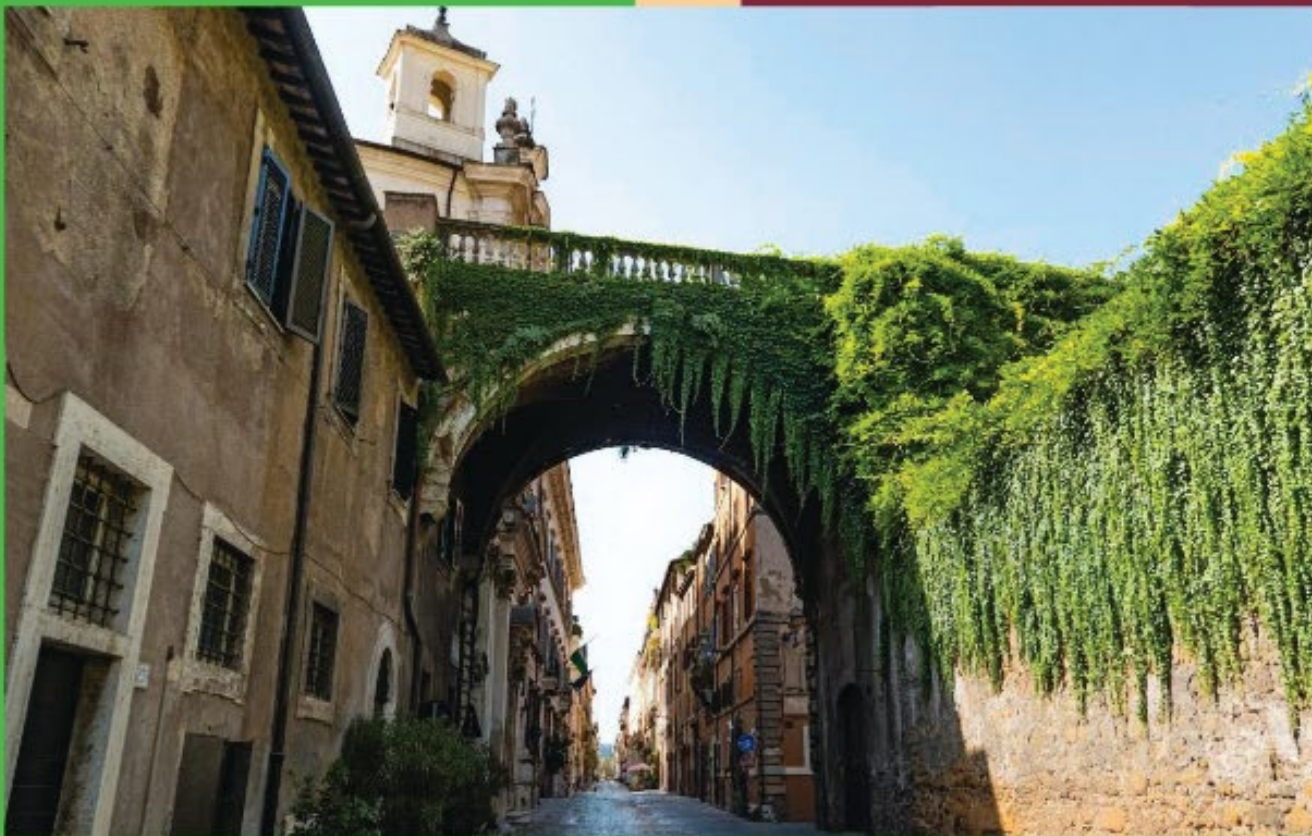


8-TALENT:
Transition Adolescence and young adults -Endocrine diseases management

An Endo-European Reference Network event

8th Conference Meeting

ROME
FEBRUARY 11.12.13
2026



FRIDAY FEBRUARY 13th

"VOICES IN TRANSITION": ROUNDTABLE BETWEEN PHYSICIANS AND PATIENT ASSOCIATIONS

Moderators: M.L. Jaffrain; M. Spaziani; F. Carlomagno

12:00-14:00 **Panel Discussion**

MTG 1: Discussant: **Rosa Maria Paragliola**
Patient Association Representative: Associazione Italiana Pazienti Addison - AIPAd (*Simone Genovese, Roma*)

MTG 2: Discussant: **Stefano Frara**
Patient Association Representative: Associazione Italiana dei Pazienti con Disordini Rari del Metabolismo del Fosfato - AIFOSF (*Nicoletta Schio, Vicenza*)

MTG 3: Discussant: **Tiziana Filardi**
Patient Association Representative: Federdiabete Lazio APS (*Lina Delle Monache, Roma*)

MTG 4: Discussant: **Franz Sesti**
Patient Association Representative: Associazione Italiana Tumori Neuroendocrini - AINET (*Giovanni Guglielmi, Roma*)

MTG 5: Discussant: **Marianna Minnetti**
Patient Association Representative: Associazione Sindrome di Bardet-Biedl Italia APS - ASBBI (*Susanna Genova, Palermo; Arianna Foti, Monza*)

MTG 6: Discussant: **Antonio Bianchi**
Patient Association Representative: Associazione Italiana Displasia Setto Ottica e altri Disordini Neuroendocrini - SOD ITALIA APS (*Diana Vitali, Roma*)

MTG 7: Discussant: **Marta Tenuta**
Patient Association Representative: Nascere Klinefelter APS (*Pamela Pedretti, Firenze*)

MTG 8: Discussant: **Carlotta Pozza**
Patient Association Representative: Comitato Associazioni Pazienti Endocrini & La Lumaca ODV (*Anna Maria Biancifiori, Perugia*)

Conclusion of the Conference

TALENT for SEX DEVELOPMENT

Moderators: M. Cappa; U. Di

09:00-09:20 **LECTURE 19:** (search for shared Holstein, Lubec

09:20-09:40 **LECTURE 20:** (the lifespan - Kr of Athens, Greece

09:40-10:00 **LECTURE EAA** steroids: puberty **Hedi Claahsen,**

10:00-10:20 Discussion

TALENT for THYROID DISEASE

Moderators: A. Pontecorvi; I

10:20-10:40 **LECTURE 22:** (strategies and c Belgium

10:40-11:00 **LECTURE 23:** (molecular-based Italy

11:00-11:20 **LECTURE 24:** (getting it wrong Hospital, Rome

11:20-11:40 Discussion

11:40-12:00 Coffee break

15:00-15:20 **LECTURE** hyperph **Bambini,**

15:20-15:40 Discussion

15:40-16:10 Coffee break

TALENT'S WORKING GROUP

9:00-10:30 C

10:30-12:00 N

12:00-13:00 L

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00-13:20 L

13:20-13:40 L

13:40-14:00 L

14:00-14:20 D

14:20-14:40 L

14:40-15:00 L

15:00-15:20 Discussion

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

TALENT CONFERENCE

13:00-13:30 Registr

13:30-14:00 Openin

TALENT for ADRENAL

Moderators: C. Scar

14:00-14:20 **LECTURE** manag **Haut-L**

14:20-14:40 **LECTURE** emergi **Karolin**

14:40-15:00 **LECTURE** Addiso **Naples**

15:00-15:20 Discuss

TALENT for DISORDERS

MTG2 Symposium

Moderators: E. Kass

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00-13:20 L

13:20-13:40 L

13:40-14:00 L

14:00-14:20 D

14:20-14:40 L

14:40-15:00 L

15:00-15:20 Discussion

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00-13:20 L

13:20-13:40 L

13:40-14:00 L

14:00-14:20 D

14:20-14:40 L

14:40-15:00 L

15:00-15:20 Discussion

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00-13:20 L

13:20-13:40 L

13:40-14:00 L

14:00-14:20 D

14:20-14:40 L

14:40-15:00 L

15:00-15:20 Discussion

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00-13:20 L

13:20-13:40 L

13:40-14:00 L

14:00-14:20 D

14:20-14:40 L

14:40-15:00 L

15:00-15:20 Discussion

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00-13:20 L

13:20-13:40 L

13:40-14:00 L

14:00-14:20 D

14:20-14:40 L

14:40-15:00 L

15:00-15:20 Discussion

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00-13:20 L

13:20-13:40 L

13:40-14:00 L

14:00-14:20 D

14:20-14:40 L

14:40-15:00 L

15:00-15:20 Discussion

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00-13:20 L

13:20-13:40 L

13:40-14:00 L

14:00-14:20 D

14:20-14:40 L

14:40-15:00 L

15:00-15:20 Discussion

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00-13:20 L

13:20-13:40 L

13:40-14:00 L

14:00-14:20 D

14:20-14:40 L

14:40-15:00 L

15:00-15:20 Discussion

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00-13:20 L

13:20-13:40 L

13:40-14:00 L

14:00-14:20 D

14:20-14:40 L

14:40-15:00 L

15:00-15:20 Discussion

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00-13:20 L

13:20-13:40 L

13:40-14:00 L

14:00-14:20 D

14:20-14:40 L

14:40-15:00 L

15:00-15:20 Discussion

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00-13:20 L

13:20-13:40 L

13:40-14:00 L

14:00-14:20 D

14:20-14:40 L

14:40-15:00 L

15:00-15:20 Discussion

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00-13:20 L

13:20-13:40 L

13:40-14:00 L

14:00-14:20 D

14:20-14:40 L

14:40-15:00 L

15:00-15:20 Discussion

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00-13:20 L

13:20-13:40 L

13:40-14:00 L

14:00-14:20 D

14:20-14:40 L

14:40-15:00 L

15:00-15:20 Discussion

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00-13:20 L

13:20-13:40 L

13:40-14:00 L

14:00-14:20 D

14:20-14:40 L

14:40-15:00 L

15:00-15:20 Discussion

15:20-15:40 **LECTURE** and skin **Bambini,**

15:40-16:00 **LECTURE** challenges **Depart** **Milan, Italy**

16:00-16:20 L

16:20-16:40 D

16:40-17:00 C

13:00



Transition of Care Working Group research activity



Mapping Transition of Care for Rare Endocrine Conditions: Findings from a Cross-Sectional Survey by the Endo-ERN ToC Working Group

Journal:	Endocrine Connections
Manuscript ID	EC-25-0798
Manuscript Type:	Research Paper
Keywords:	transition, rare endocrine conditions (RECs), transitional care, Rare diseases/syndromes, Endo-ERN



Under review in *Endocrine connections*



Co-operation – ESE-ESPE Transition of Care Working Group

**European Society for Paediatric Endocrinology (ESPE)
and European Society of Endocrinology (ESE)
Joint Clinical Practice Guidance for Healthcare
Transition from Paediatric to Adult Endocrine Care**
*(Accepted for publication by the European Journal of
Endocrinology (EJE) and Hormone Research in
Paediatrics (HRP))*

Next steps in discussion...:

- Joint educational activities?
- Research?

Co-operation – Overarching ERN Transition of Care Working Group

- Aim: to create a coordinating group for all transition-related work across ERNs
- Development of official structure and mandate as requested by EU
- Core group

Position	Name	ERN
Coordinator	Ruth De Bruyne	ERN RARE LIVER
Paediatric lead	Ramon Gorter	ERNICA
	Raffaella Colombatti and Anna Collado	ERN EuroBloodNet
Adult lead	Karolina Stepien	METAB ERN
	Katarzyna Nowomiejska	ERN EYE
Paramedic	Paloma Martinez	ERN transplant child
	Guillermo M. Sibón Sancho	ERN EYE
Geneticist	Liesbeth Wildero Van Wouwe	VASCERN
ePAG	Karsten Vanden Wyngaert	ERKNET
Youth panel member	Bent Hennersdorf	ERN RARE LIVER
	TBD	TBD
Junior researcher	Rebecca Pulvirenti	ERN ERNICA
	Janne Suykens	ERN RARE LIVER

- Recruitment of Youth panel
- Regular meetings of plenary working group

Co-operation – Overarching ERN Transition of Care Working Group



Endo-ERN Representatives:

- Ulla Döhnert (paediatric endocrinology)
- Claudio Auteri (patient representative)

Transversal Working Group on Research

Alberto Pereira, Adult Chair Endo-ERN

Olaf Hiort, Paediatric Chair Endo-ERN

Charlotte van Beuzekom, Project Manager Endo-ERN



Funded by
the European Union



European Journal of Endocrinology, 2025, 193, ii128–ii131
<https://doi.org/10.1093/ejendo/lvaf072>

Review Article



EndoCompass project: rare diseases in endocrinology

Alberto M. Pereira^{1,2,*} and Olaf Hiort^{2,3}

¹Department of Endocrinology and Metabolism, Amsterdam University Medical Center, Meibergdreef 9, 1105AZ Amsterdam, the Netherlands

²European Reference Network on Rare Endocrine Conditions (Endo-ERN)

³Sektion für Pädiatrische Endokrinologie und Diabetologie, Klinik für Kinder- und Jugendmedizin, Universität zu Lübeck, Ratzeburger Allee 160, Lübeck 23562, Germany

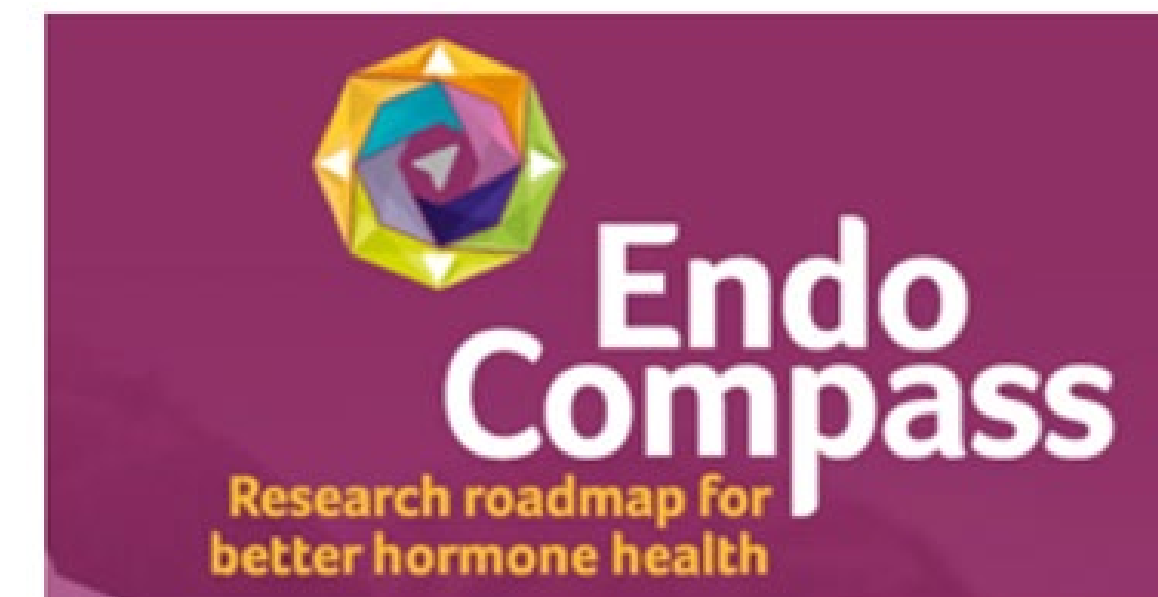
*Corresponding author: Department of Endocrinology and Metabolism, Amsterdam University Medical Center, Meibergdreef 9, 1105AZ Amsterdam, the Netherlands. Email: a.m.pereiraarias@amsterdamumc.nl

- Digital tool and pathway development
 - Develop/implement disease-specific clinical outcome assessment tools
4. Innovative therapies:
 - Advanced therapeutic medicinal products need assessment/mapping in Endo-ERN
 - Analysis of specific therapy and treatment availability across EU member states
 5. Unique clinical resource development/pilot:
 - Disease-specific patient journey development
 - Rare endocrine disease patient pathway development/application
 - Mapping of European patient referral pathways/channels in Endo-ERN

Focus areas for research into rare conditions

The following are examples of specific topics that should be considered when deciding on a research grant. They have been incorporated into the Endo-ERN Quality of Care Improvement and Research Counsel:

1. Diagnostic research readiness:
 - Assessment of data readiness, standardization, and harmonization
 - Genome re-analysis pipeline application, variant interpretation, and translation to endocrinology departments or clinics
 - Genomic innovation to shorten time to diagnosis
2. Clinical trial readiness:
 - Multinational patient inclusion and collaboration
 - Disease progression modelling
3. Clinical outcome assessment:
 - Estimating the socio-economic impact of rare endocrine diseases



Research WG

Implement ***Focus areas for research into rare conditions***

- Map research landscape within Endo-ERN and the wider ERN ecosystem
- Raise awareness of persistent underfunding of endocrine research despite its major impact on the EU population
- Highlight patient impact and unmet needs in rare endocrine conditions

Other RD Project updates

Charlotte van Beuzekom, Project Manager Endo-ERN



Funded by
the European Union

Other RD project updates



European
Reference
Network



Endo-ERN

ERDERA

European **Rare Diseases**
Research Alliance

ERDERA

JTC 2026

- Resolving unsolved cases in rare genetic and non-genetic diseases through variant validation and new technological approaches
- Proposal initiatives:
 1. Martin Kircher (Luebeck): DSD
 2. Luca Persani (Milan): Rare Thyroid Disorders
 3. Pauline Romanet (Marseille): MEN1
 4. Marco Bonomi (Milan): CHH

Networking Support Scheme

- Funds for events that promote RD knowledge sharing
- Max Euro 30.000 per event
- Constant open call, next deadline 7 April 2026

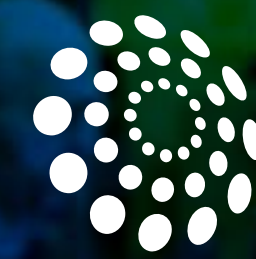
www.erdera.org

Realised

- Factors influencing clinical trial enrolment in rare and ultra-rare diseases
- 👉 Open until 28 January 2026 <https://forms.office.com/e/nhaqB5gvN6>

AOB & questions





European
Reference
Network



Endo-ERN