

Endo-ERN Steering Committee / Advisory Board meeting

Date: Tuesday 2 September 2025

Time: 09:30 – 11:30 hrs



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the European Union

Agenda

1. Work Package updates (8 minutes each)
2. Working Groups updates (8 minutes each)
3. Other RD project updates
4. AOB & questions

WP1 Management & Coordination

Alberto Pereira, Adult Chair Endo-ERN

Olaf Hiort, Paediatric Chair Endo-ERN

Charlotte van Beuzekom, Project Manager Endo-ERN



Personnel communication



- Jette Kristensen



- Arlene Smyth



- Laura Maag



- Marc Büdenbender



Funding grants



Current EU4Health grant 2023-2027

- milestones & deliverables
- interim report, Feb 2026

Approved deliverables

Nr.	Name
D1.1	ERN Development Plan
D1.2	Structural Map for Endo-ERN
D1.3	Organisation General Assembly Y1
D2.1	Dissemination Plan
D3.1	Evaluation Plan
D3.4	Indicators report Y1

Multiannual Financial Framework 2028-2034

- absence of stand-alone successor to EU4Health program
- joined forces, aiming to sustain operating ERN grants

Upcoming 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



Lead author	Paper title	Submitted	Deadline
Felix Reschke	Challenges and Strategies for Transitioning Adolescents with Rare Forms of Diabetes to Adult Care: Perspectives and Evidence-Based Approaches	Yes	
Nienke Biermasz	Quality of care for patients with prolactinoma diagnosed under the age of 25 years		September 30th
Hedi Claahsen	Enhancing Transition Care for adolescents and young adults with adrenal insufficiency in the Netherlands: A Holistic Model for Improved Patient Outcomes	Yes, revised paper	
Corinna Grasemann	Continuity of Care in Klinefelter Syndrome: Age-Adapted Modules for Standardized Clinical Data Collection (I-KS)	Yes, revised paper	
Violeta Iotova	Feasibility of transition research in pituitary disease using patient registries		August 8th
Ana Priego	The EuRRECa Core Registry and its potential for studying transition of care in Endo-ERN reference centres		July 15th
Emily White	First 5 Years of Endo-ERN: Evaluation of the Network and Member Expert Centres & Creation of Endo-ERN Performance System		September 15th
	Acromegaly: The Patient Journey		September 15th
Judit Toke	Transitional care of patients with congenital adrenal hyperplasia - our experiences and challenges in a tertiary adult referral center in Hungary	Yes	
George Mastorakos	TRANSITION of pediatric patients on GH treatment in Greece		September 1st
Alberto Pereira / Olaf Hlort	Introduction		

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

Various

Endo-ERN Research Working Group

- chairs
- members

Call for new Affiliated Partners

GA 2026

- Monday 20 and Tuesday 21 April 2026
- Crown Plaza Hotel Athens, Greece
- Program - Invitations

MTG HCP meetings

MTG	Date	Time
1		
2	10 October 2025	15:00 hrs
3	20 October 2025	16-00 hrs
4		
5	08 September 2025	15:00 hrs
6	14 November 2025	11:00 hrs
7	15 October 2025	16:00 hrs
8	27 October 2025	15:00 hrs

WP2 Communication & Dissemination

Aimee Casey, Communication Manager, Endo-ERN

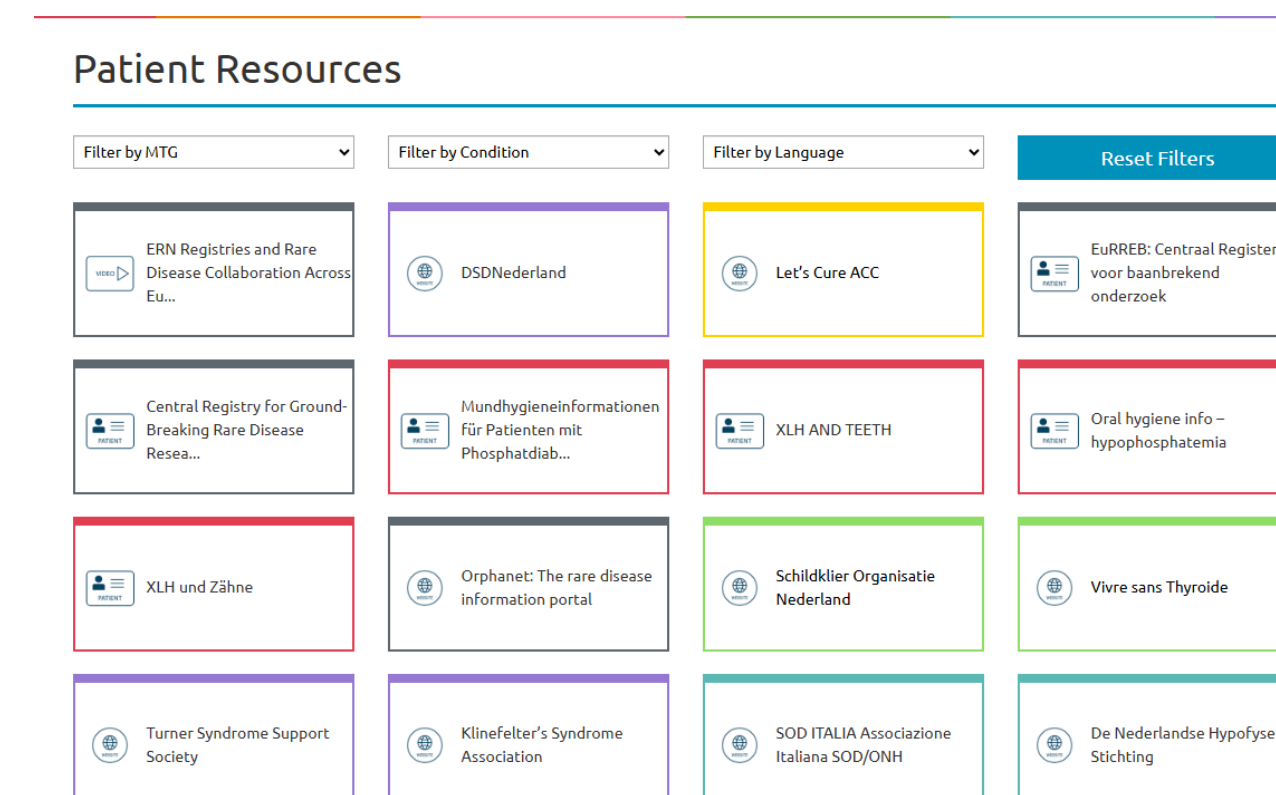


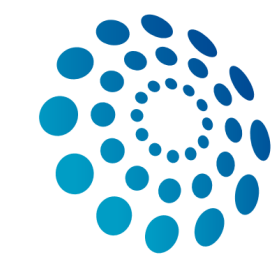
Social media update
2025

	Current total	Increase from May
LinkedIn	732	+69
Facebook	456	+21
YouTube	644	+52
Newsletter subs	2462	+30

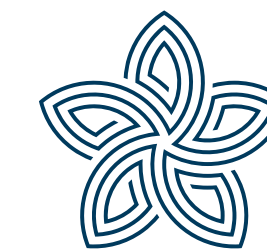
Patient Resources

- Over 100 searchable patient resources including links, documents and videos
- AI generated summary of each item in English and item language – increase searchability
- Continue to populate with resources with the support of ePAGs and MTG chairs
- Dedicated promotion on Facebook





European
Reference
Network



Endo-ERN

Newsletters

- Topic based in effect from September
- ***Digest*** for primary and sub representatives – first Friday of the month



Autumn Activity



National Association Outreach

- Database complete – identification of national coordinators and validation
- Landing page in development

Commission Continuous Monitoring Report

- Expected end of September will underpin ERN-wide awareness campaign
- Phase 2 showcase Endo-ERN using performance data

Ongoing promotion

- Webinars, exchange programme, etc

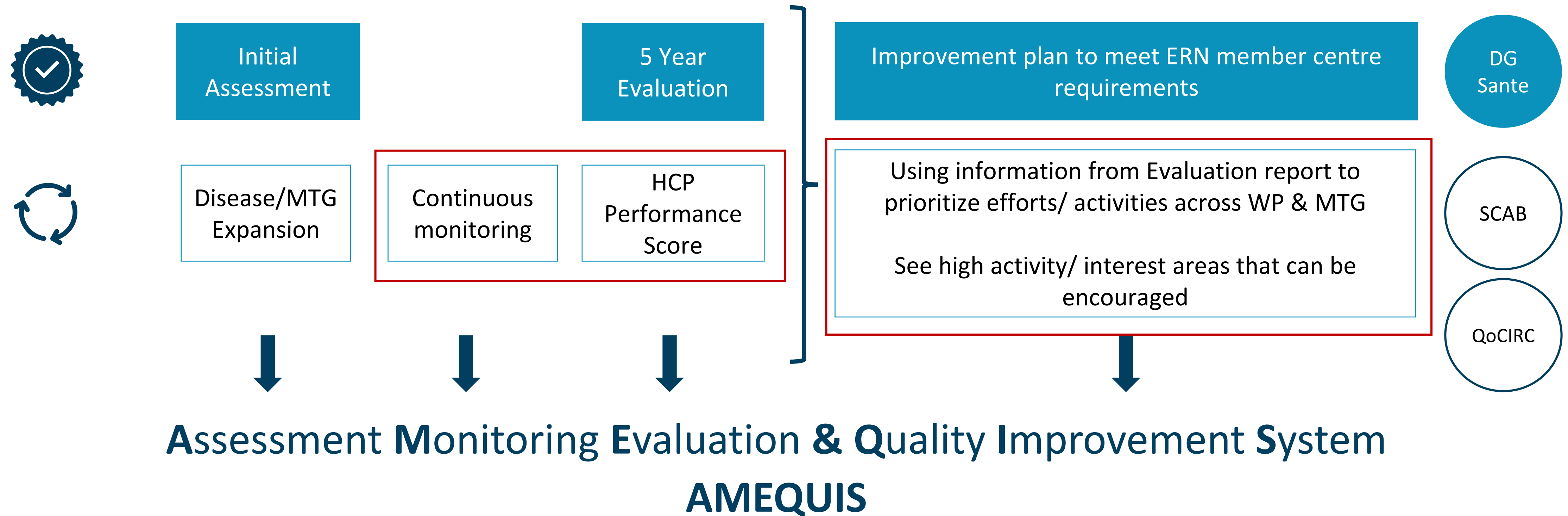
WP3 Evaluation

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



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Evaluation in Endo-ERN – QI



Continuous Monitoring

Data from 2023 & 2024

DG Sante Report to be published in September of aggregated data for all ERNs

Exercise in 2025

2025 data collection starting from January 2026

Key reminders to all members:

- Update your e-REC prior to end of the year
- Request reports of total numbers from your administration in time (if necessary)

Continuous Monitoring Endo-ERN

Who

- All Endo-ERN member HCPs
- All Affiliated Partners

What

- Measure of impact ERN has in our member HCPs
- Check HCPs complete membership activities for Endo-ERN
- **Mandatory** exercise from European Commission
- Signature Annual Conflict of Interest

How

- Record data based on 18 Indicators via Survey from Endo-ERN
- Regular E-REC data submission
- Report totals to European commission directly



When

- Annually – once a year in the first quarter
- **2025 data collected starting from January 2026**

Outcomes

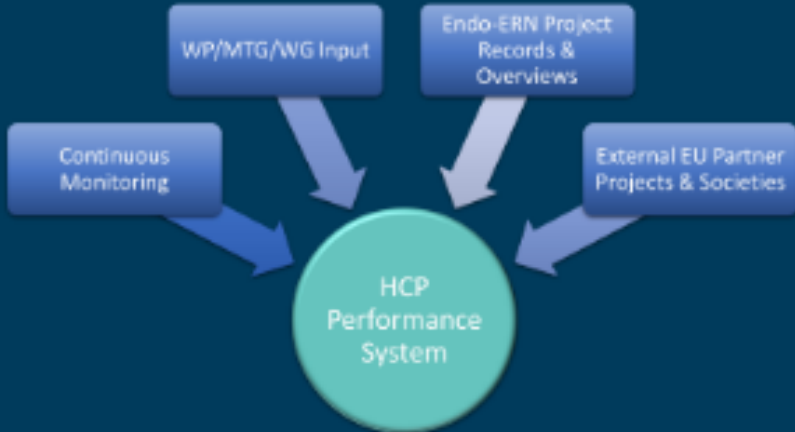
- HCP performance system – internal scoring
- Changes to evaluation processes
- Map & Showcase ongoing efforts of our HCPs

*Please add
this overview
slide to your
MTG HCP
meetings*

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/ hosted over 300 rare disease specific training and educational activities
- Used the 67 guidelines and CDSTs endorsed by Endo-ERN and highlighted 6 more worth endorsement
- Attended and contributed 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 activities a HCP is scored on. These scores are weighted based on their category.

Possible Points


21 pts mandatory = 60% of total score	The resulting weighted values are used to calculate the overall Overall Score. Please see our website for further details.
20 pts expected = 25% of total score	
22 pts bonus = 15% of total score	

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


Expected Activities = not mandated for membership but expected of contributing member HCPs (max. 2 points per activity)

Bonus Activities = (roles) undertaken by HCPs/ individual experts therein that advances our mission and seeks to improve the Endo-ERN (1-3 points per activity)

Disclaimer: The scores generated within the Endo-ERN HCP Performance System are intended to measure the contribution and participation in activities as a member of our network. Please note these scores do not assess or reflect in any way adherence to standards of clinical care (nationally or locally).



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HCP Performance Reports 2024

(Later this year due to edits in ERN monitoring report reveiws)

All HCPs in Endo-ERN will receive details to access their reports this week and have opportunity to comment/correct any errors.

Deadline for comments = **19th September 2025**

HCP Resources Mapping

To update our information and direct efforts in the network to the correct audience

WP / WG	What is collected
Genetic Testing WG	Genetic testing labs linked to Endo-ERN HCPs Diagnosis expertise & tests per lab
WP4 – CPMS 2.0 Permission & Expertise	Permission to use system (Privacy/DPO) Ability to use system in HCP (ICT) Expertise in your HCP – frequently sought professionals in system e.g. paediatric neurosurgeon
WP1 – Rare disease office	Is it present in your HCP? National network discussions regularly held? Cross border care agreements present (y/n) with specific countries
WP8/9 - links with Ukraine	Do you receive referrals from Ukraine HCPs DO you have a policy on offering care to patients from Ukraine (still) in place?

Evaluation WG



Individual Options/Recommendations for evaluation and on-site audit **structure improvement:**

Within the 5 year evaluation WG Endo-ERN has provided 5 key change recommendations (with thanks to our HCPs who contributed):

1. Remove interview with hospital patients
2. Minimise time commitment
3. Eliminate redundancies and duplicated work
4. Utilise existing evaluations
5. Refine MEs criteria for online submission

WP4 Healthcare & CPMS

Olaf Hiort, Paediatric Chair Endo-ERN

Greta Ginski, CPMS Operational Helpdesk Manager

Emily White, Project Manager & CPMS Operational Helpdesk Manager

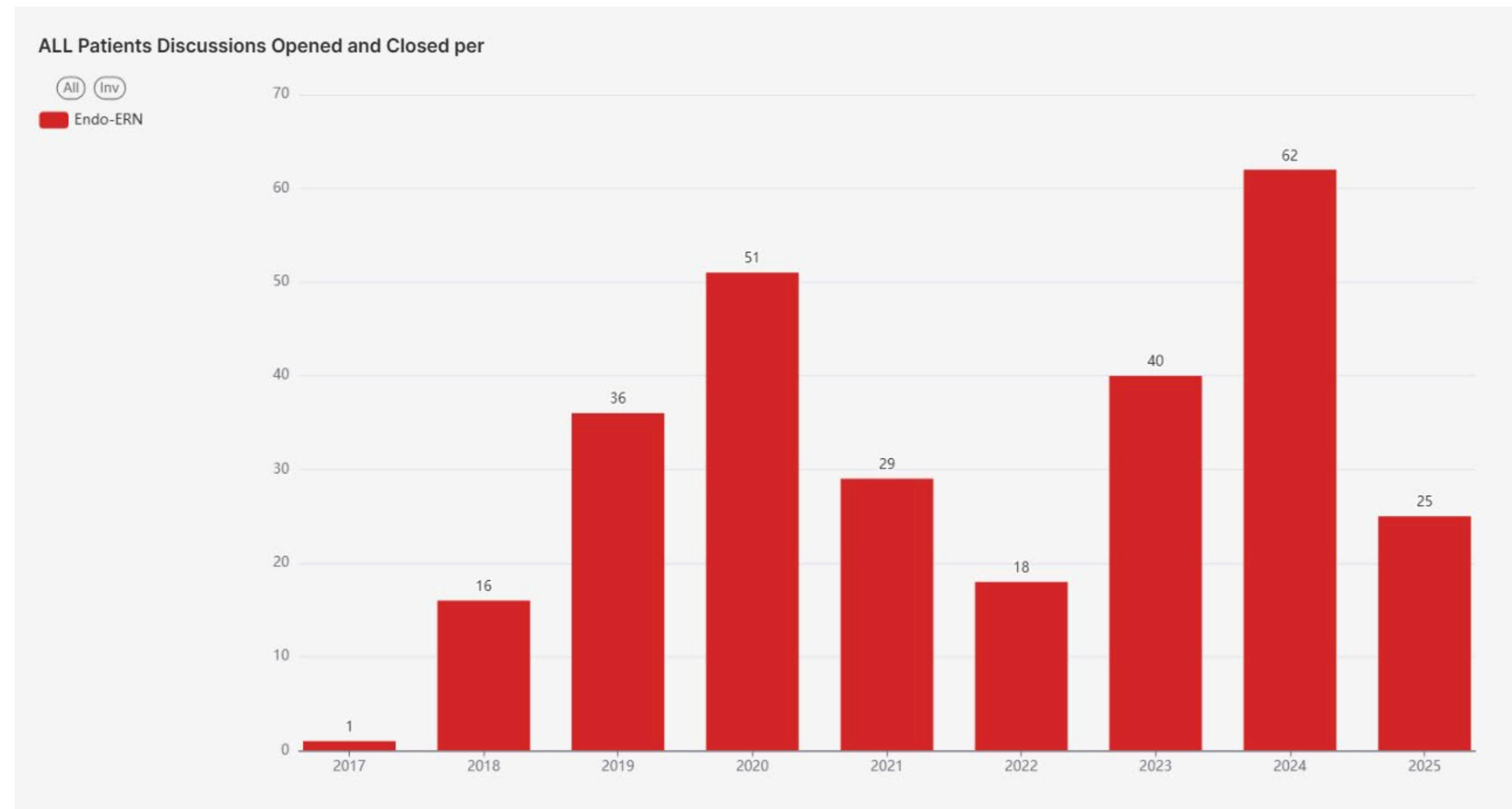


Current Statistics



- Active users in 2025= 96 (60 in CPMS 2.0 **only**)
- 358 user roles
- 942 patients enrolled

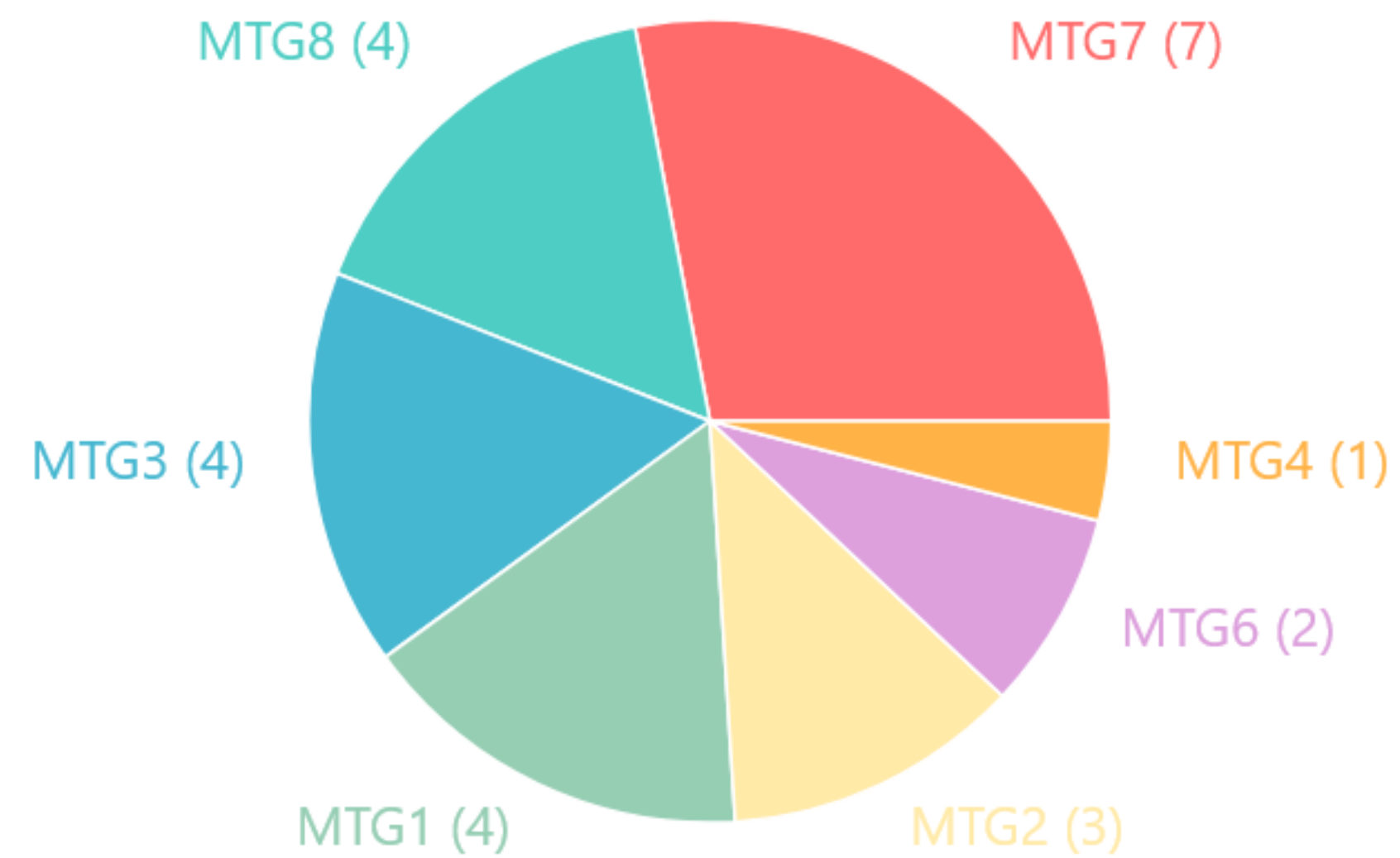
Note:
Discussion = 'Panels'



Enrolled patient cases by MTG

CPMS 2.0 Patient Cases Distribution per MTG

Total cases: 25



Patient cases by HCP and Country



Healthcare Provider	Country	Cases
AOU Meyer -- Florence	Italy	4
Radboud University Medical Centre Nijmegen	Netherlands	3
Universitätsklinikum Schleswig-Holstein	Germany	2
Leiden University Medical Center	Netherlands	2
Hôpital Universitaire de Bruxelles (HUDERF-Erasme)	Belgium	1
Aarhus University Hospital	Denmark	1
AOU -- Modena	Italy	1
General Hospital of Athens LAIKO	Greece	1

AO City of Health and Science -- Turin	Italy	1
Consortium: Katholisches Klinikum Bochum	Germany	1
MHAT Sveta Marina	Bulgaria	1
HCP Cluj County Emergency Hospital	Romania	1
Erasmus MC: University Medical Center Rotterdam	Netherlands	1
Maria Sklodowska-Curie National Research Institute of Oncology	Poland	1
Foundation IRCCS CA'Granda Ospedale Maggiore polyclinic -- Milan	Italy	1
Children's Clinical University Hospital, Riga	Latvia	1
Amsterdam University Medical Centers Location AMC	Netherlands	1

37 different HCPs from 15 different countries have attended meetings in CPMS 2.0




61 experts have attended a meeting in CPMS 2.0 (excluding Teams/Webex calls)

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 Skłodowska-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

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



CPMS 2.0 Goes Mobile!

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



Available in



Download the CPMS 2.0 App now

Go to the app store and download the CPMS 2.0 App (scan code below)

Login with your EU Login details and use the EU Login App to authenticate yourself –
just like in the desktop version



New MTG meeting structure



Proposal for structured agenda in CPMS 2.0 Meetings

- Main agenda point
 - General care discussion
 - Thematic case collection around a specific disease or clinical question
 - Presentation of particularly interesting or instructive cases
- Slots for case discussions

Rescheduling of MTG meeting series in CPMS 2.0

- See meetings section in CPMS 2.0 for the meetings you are invited to
- Check website for accurate dates and times

MTG meeting status

MTG	Next date	Main agenda point?	Case scheduled?
MTG 1	1st Oct @15:00		LSmo (follow-up discussion)?
MTG2	TBD		
MTG3	24th Sep @13:30		
MTG4	TBD		
MTG5	1st Sep @15:00	Case of hypopituitarism	
MTG6	19th Sep @13:00		CushingC?
MTG7	3rd Sep @16:00	Care pathways in CAH	
MTG8	1st Oct @9:00	Care discussion treatment with β -blockers or Triac in RTH β patients	Myanna2? Ramona Curticaean? Leonardo Xenon 705

New website resources



Short videos

Go visit the [Endo-ERN website](#) to check out new training materials including snippet videos on how to use the new app!

Ukraine patients

Ukrainian clinicians can use CPMS 2.0 for patient case consultations
Coordination will be handled via the hub

New website resources



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



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Endo-ERN members activity

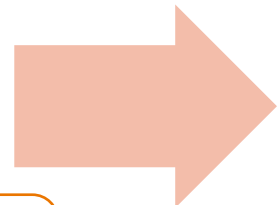


e-REC active centres

Grant indicator:
75% by 2027

December 2024

- 87 centres (82%)



August 2025

- 94 centres (90%)

New cases in 2025

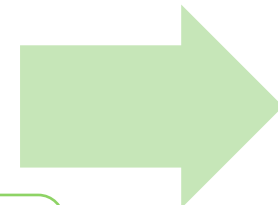
9635

Core Registry centres with cases

Grant indicator:
50% by 2027

December 2024

- 31 centres (29%)



August 2025

- 40 centres (38%)

630



Key Performance Indicators (KPIs) planning

- In collaboration with Coordination **proposed KPI 1:**
 - Time to diagnosis in the expert center
- **Proposed KPI 2:**
 - e-REC to CORE ratio



Study Groups and Working Groups

New Module

- MEN 1 (in testing phase)

New Working Groups

- Pregnancy and fertility (module beta-version in testing phase)
- Hypoparathyroidism (in starting phase)



NEW: MEN 1 module



Basic module

Genetic testing

Follow up

Patient history

Assessment Date

?

yyyy-mm-dd

Type of diagnosis

?

☐ Predictive testing as child of an affected parent

☐ Cascade screening because a new diagnosis of MEN1 in a (first-degree) relative

☐ Genetic testing because of clinical phenotype or a clinical/familial diagnosis

☐ Clinical diagnosis (2 MEN1 main tumors)

☐ One MEN1 main tumor plus a diagnosis of MEN1 in first-degree relative(s)

MEN1 affected first-degree relatives

?

☐ Mother

☐ Father

☐ Brother(s)

☐ Sister(s)

☐ Children

First MEN1-related clinical manifestation

?

☐ Primary hyperparathyroidism

☐ Pituitary adenoma

☐ DP neuroendocrine tumours (NET)

☐ Lung NET

☐ Thymus NET

☐ Gastric NET

☐ Distant metastasis from a NET

☐ Adrenal lesions

☐ Skin lesions

☐ Other manifestations

Clinical manifestation

Has the patient ever been diagnosed with...?

?

☐ Primary hyperparathyroidism

☐ Pituitary adenoma

☐ DP neuroendocrine tumours (NET)

☐ Lung NET

☐ Thymus NET

☐ Gastric NET

☐ Distant metastasis from a NET

☐ Adrenal lesions

☐ Skin lesions

☐ Other manifestations

EuRREB symposium



EuRREB Symposium		
12 September 2025		
Academy Building, Rapenburg 73, Leiden, the Netherlands		
09:30 - 10:00	Registration - Welcome coffee and tea	
10:00 - 10:05	Opening	Natasha Appelman-Dijkstra
10:05 - 10:25	Registries and ERN's	Luca Sangiorgi
10:25 - 10:45	5 years of European Registries of Rare Endocrine and Bone Conditions	Ana Priego Zurita
10:45 - 10:55	The Melorheostosis module: a collaboration between ECTS, ERN BOND and EuRREB	Martine Cohen Solal
10:55 - 11:05	The LCH module: a collaboration between ERN BOND, ERN-EuroBloodNet and Endo-ERN	Polyzois Makras
11:05 - 11:15	Outcomes of the FD/MAS module	Sophie Huisman
11:15 - 11:25	The Rare Hypophosphataemia module	Alexandra Ertl
11:25 - 11:35	Contributing to the Achondroplasia module: a student experience	Chady Omara
11:35 - 11:45	The importance of a pregnancy module	Claudia Finis
11:45 - 12:05	Discussion: How to proceed with the first modules?	Facilitated by Faisal Ahmed
12:05 - 12:25	Coffee break	
12:25 - 12:45	Remodeling an existing rare disease registry to be used in regulatory context	Marina Mordenti
12:45 - 13:00	Ethics approval and data sharing process	Tess de Rooij
13:00 - 13:20	FAIR Data Stewardship	Bruna Dos Santos Vieira
13:20 - 13:40	Rare Disease Registries – seeing the forest for the trees	Faisal Ahmed
13:40 - 13:50	Closing words and group picture	Faisal Ahmed/Tess de Rooij



WP6 Education and Training

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



Endorsements



- 12 endorsed events for 2025 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

- 6 Approved Applicants for 2025 (3/6 completed)
- Call for 2026 will be open from October 1 until November 30
- Possible prolonging of the exchange period in process

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

Webinars per MTG for 2025 so far

MTG1	3 Webinars
MTG 2	1 Webinar
MTG3	0 Webinars
MTG4	0 Webinars
MTG5	2 Webinars
MTG6	4 Webinars
MTG7	4 Webinars
MTG8	1 Webinar

Accreditation

- Accreditation means more registrants and attendees
- Minimum 5 webinars per accreditation pack
- 1 more webinar required to complete the yearly target from the EU Commision

Upcoming Webinars 2025



MTG	Title	Date
2	Clinical Practice recommendations for the diagnosis and management of XLH	November 4,2025
5	An update on primordial dwarfism conditions	October 20,2025
8	Diagnosis and management of TSH resistance	October 7,2025
1	Genotype-phenotype correlations in CAH.	September 24,2025
1	Advances in the diagnosis and management of Congenital Hypogonadotropic Hypogonadism (CHH)	September 22,2025

←

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

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European
Reference
Network



Endo-ERN

Rare Endocrine Conditions ▾

About Us ▾

For Professionals ▾

For Members ▾

For Patients ▾

🔍

🏠 / Webinars

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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				
	Webinar on the Clinical Practice recommendations for the diagnosis and management of XLH	2025-10-29	17:00 – 18:00 hr				
	An update on primordial dwarfism conditions webinar	2025-10-20	15:00 – 16:00 hr				
	Diagnosis and management of TSH resistance	2025-10-07	17:00 – 18:00 hr	Edward Visser and Luca Persani			
	Genotype-phenotype correlations in CAH	2025-09-24	16:00 – 17:00 hr				
	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				
	Congenital combined pituitary hormone deficiency in Europe webinar	2025-06-25	16:00 – 17:00 hr				
	Cushing's Syndrome Webinar	2025-06-10	16:00 – 18:00 hr	Jérôme Bertherat, Marta Baranska and			

WP7 Clinical Practice Guidelines & Clinical Decision Support Tools

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



Work package members

Anna Nordenström [Paediatric chair](#)
Karolinska University Hospital (Stockholm, Sweden)



Olaf Dekkers [Adult chair](#)
Leiden University Medical Center (Leiden , Netherlands)



Jette Kristensen [ePAG representative](#)
Addison Foreningen Danmark (Denmark)



Alessandro Lazzerini [ePAG representative](#)
Spanish Association of Congenital Adrenal Hyperplasia (CAH) (Spain)



Diana Vitali [ePAG representative](#)
SOD ITALIA – Italian organization for septo optic dysplasia and other neuroendocrine disorders (Roma, Italy)



Dr. Charlotte Gaasterland Guideline methodologist
LUMC, department of Epidemiology



Main goal WP 7



Main goal:

- ☐ To develop EndoERN clinical practice guidelines and decision-making tools for identified subject areas. Focus on identifying knowledge gaps and guideline needs.

- ☐ Key considerations include:
 - Covering the transition from pediatric to adult endocrinology and
 - Ensuring alignment with EndoERN's core objectives
 - Patient perspectives and needs will be central to the guideline development process,
 - Patients have an active involvement.

- ☐ The impact of the guidelines will be assessed, and methodological support provided.

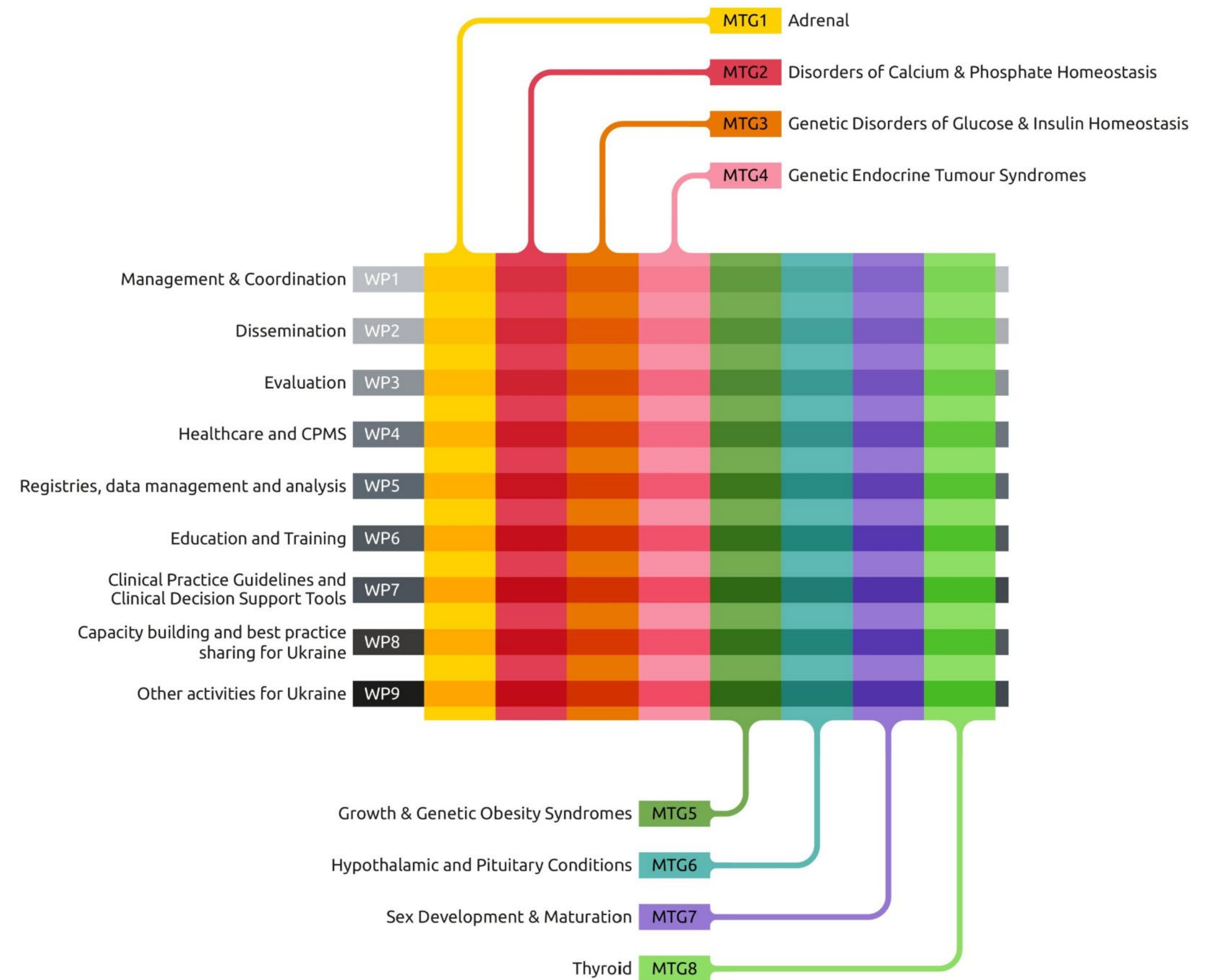
- ☐ A previous guideline on puberty treatment was published in 2022 and has received significant citations.

Collaboration

Work Packages 2 and 6

Vital for:

- Education
 - Training, and
 - Guideline dissemination efforts.
- ☐ Development of infographics for clear communication of key guideline messages.
 - ☐ Standardized presentation of guidelines will be pursued to enhance recognition.



Endo-ERN Guidelines

Pubertal induction and transition to adult sex hormone replacement , Published April 2022 J Journal Endocrinology

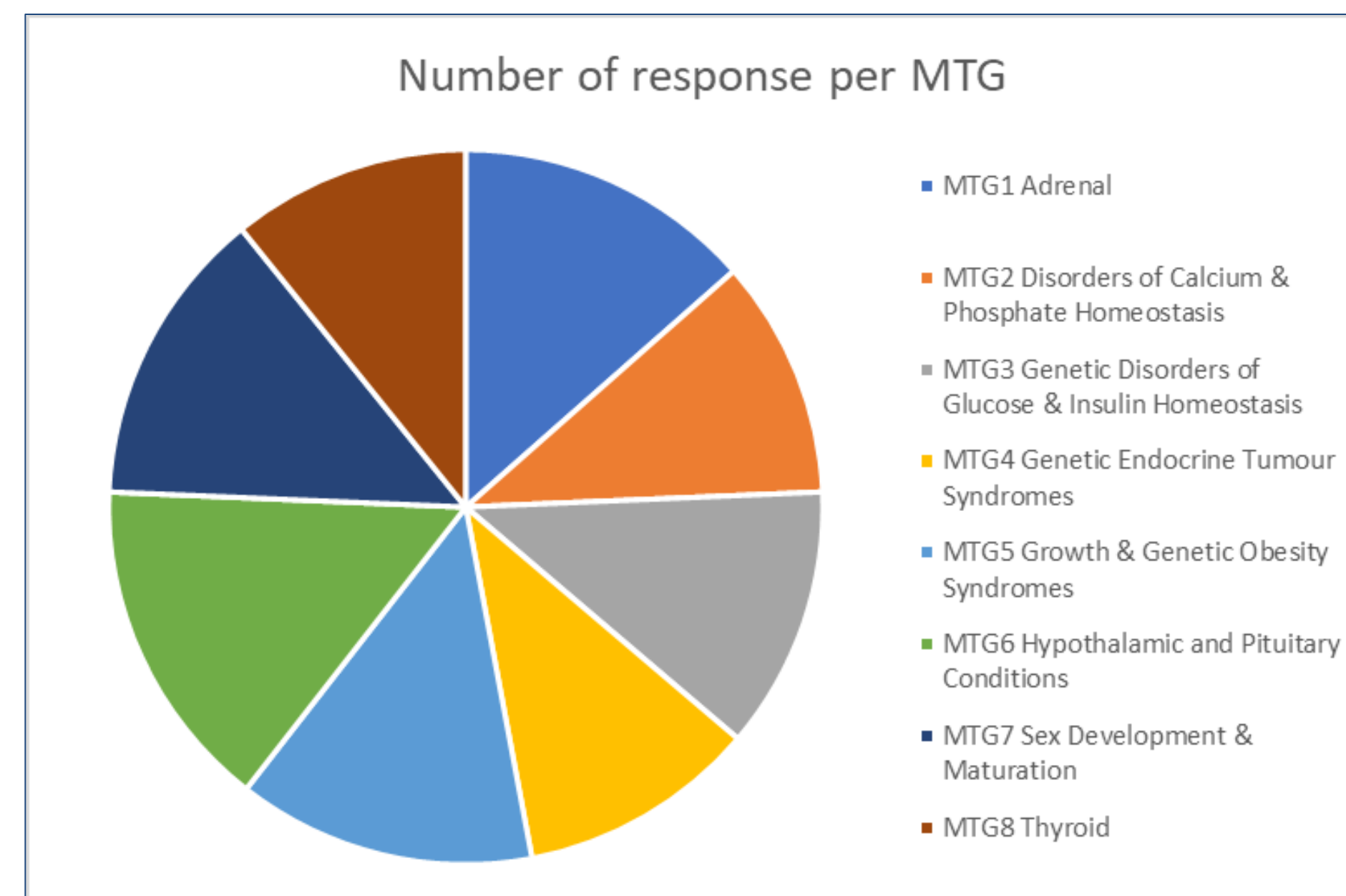
Familial Hyperaldosteronism, Published April 2024, presented ECE May 2024

Transition in collaboration with MTG 6, initiated

Next CPG topics

- WP7 asks all members for theme suggestions in a survey
- WP7 will comment on the feasibility of the topics from a methodological point of view
- These topics plus comments will be discussed with the Steering Committee, who will then make a decision

Survey



Top voted topics (**exclud.** new suggestions):

18 votes - Hypothalamic obesity treatment and management (of craniopharyngioma) in children and adults (MTG5/6)

14 votes - Replacement treatment in hypoadrenalism: focus on monitoring target (MTG1)

14 votes - Diagnosis and Lifelong Management of Congenital Pituitary Deficiencies (MTG6)

11 votes - Prader Willi (MTG5)

Suggestion



- Start with two topics in parallel

Publish at least one guideline per year in years 2-4

Assess impact?
Downloading
Referenced



Replacement treatment in hypoadrenalism: focus on monitoring target (MTG1)



Hypothalamic obesity treatment and management (of craniopharyngioma) in children and adults (MTG5/6)

Standard Operating Procedures for the development process, methodology, and endorsement of CPGs

- SOP on guideline process & methodology is drafted
- SOP on endorsement is still in development
- No clear strategy in other ERNs – we are currently building a network with other ERN methodologists to discuss this issue

MTG specific CDSTs

Clinical Decision Support Tools



For discussion:

Lay versions, how and
what format?

Patient involvement.

- Translate
guidelines into
multiple languages

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)



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Status



Currently 24 participants from 8 different countries

Virtual meetings: bimonthly

ESHG – ERN collaboration/mutual endorsement: nomination of communication managers (E. White) and liaison managers (TE, DP (provisionally until formal vote)), first ESHG-liaison manager meeting on Sept 2nd/9th 2025

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
University Hospital	Germany	Aachen	Thomas Eggermann, Miriam Elbracht	ISO 15189	(MS)MLPA, Mi	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 turn	Abnormal growth, Imprinting Defects (BWS			32462478, 35	PMID: 332720	yes

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

- Author teams are working on the different topics

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)



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Endo-ERN 'Transition of Care' Working Group composition

N	MTG / Role	Representative	Ped/Adu	E-mail (transitionwg@endo-ern.eu)
1	Chair	Andrea Isidori	Adu	andrea.isidori@uniroma1.it
2	Co-chair	Ulla Döhnert	Ped	u.doehnert@uni-luebeck.de
3	1/Adrenal	Svetlana Lajic	Ped	svetlana.lajic@ki.se
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8	2/Calcium&Phosphate	Graziamaria Ubertini	Ped	graziamaria.ubertini@opbg.net
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10	3/Glucose&Insulin	Pietro Maffei	Adu	pietro.maffei@aopd.veneto.it
11	4/Genetic tumours	Annemarie Verrijn-Stuart	-	a.a.verrijnstuart@umcutrecht.nl
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New entry 26	ePAG	Giorgio Dal Maso	-	giorgio.dalmaso@airisc.org



European
Reference
Network



Endo-ERN

Transition of Care Working Group activities

1. Completed (as partly reported at Endo-ERN GA)

- Group formation and approval
- Literature research
- Bilateral coordination meetings with sister ERNs
- 7th TALENT meeting in Rome
- Development of ToC e-Survey, data analysis and draft of manuscript (submission in progress)

2. In progress

- Development of checklists for transition of patients with defined conditions
- ERN Overarching transition group activities
- ESE / ESPE transition coordination meetings

3. Organization of the 8th TALENT Conference meeting

- 11-13th February 2026 in Rome, Italy

Checklist for Transitioning Patients with Congenital Hypogonadotropic Hypogonadism (cHH)
(To be completed by the referring paediatric endocrinologist and shared with the receiving adult endocrinologist)

1 Patient Information

- Name & Surname: _____
- Date of Birth: ____ / ____ / ____ Gender: ☐ Male ☐ Female

2 Essential Information for the Adult Endocrinologist

- Diagnosis of cHH confirmed at age: _____
- GnRH (or dynamic) testing: ☐ Yes (☐ Attached) ☐ No
- Presence of cryptorchidism: ☐ Yes ☐ No
 - If yes, side and treatment: _____
- Olfactory function assessment performed: ☐ Yes ☐ No
 - If yes, anosmia / hyposmia / normal: _____
- Genetic analysis performed: ☐ Yes (☐ Attached) ☐ No
 - If yes, result: _____
- MRI of the CNS performed: ☐ Yes (☐ Attached) ☐ No
 - If yes, result: _____
- Bone health assessed (DXA, vertebral morphometry): ☐ Yes (☐ Attached) ☐ No
 - If yes, result: _____
- Any psychological / psychosocial issues: ☐ Yes (☐ Attached) ☐ No
 - If yes, specify: _____
- Pubertal induction therapy
 - Age at initiation: _____
 - Regimen: _____
 - Duration: _____
- Growth
 - Growth chart: ☐ Yes, attached ☐ No
 - Completed linear growth (<2 cm/year): ☐ Yes ☐ No
- Pubertal stage (according to Tanner): _____
- Testosterone (T) / Estrogen-Progestin (EP) replacement therapy
 - Current formulation: _____
 - Dose & route of administration: _____
- Semen analysis / cryopreservation performed: ☐ Yes (☐ Attached) ☐ No
 - If yes, specify: _____
 - Last evaluation date: ____ / ____ / ____ (☐ Attached)
- Presence of associated pituitary deficiencies: ☐ Yes ☐ No
 - If yes, specify: _____
 - Current therapy: _____
- Reproductive goals discussed with patient: ☐ Yes ☐ No
 - If yes, specify: _____

3 Crucial Monitoring Parameters

- Sex hormone levels (T / E2), liver function tests, complete blood count
 - Last evaluation date: ____ / ____ / ____ (☐ Attached)
- Ultrasound: Testicular volume / uterine morphometry:
 - Last evaluation date: ____ / ____ / ____ (☐ Attached)
- Bone age:
 - Last evaluation date: ____ / ____ / ____ (☐ Attached)
- Bone health (DXA scan):
 - Last evaluation date: ____ / ____ / ____ (☐ Attached)

4 Adolescent-Specific Considerations

- Counselling on smoking impact on fertility potential provided: ☐ Yes ☐ No
- Concerns regarding body image and mental health addressed: ☐ Yes ☐ No
- Sexual health counselling provided: ☐ Yes ☐ No
- Timing of first semen analysis (if applicable) discussed: ☐ Yes ☐ No
 - If yes, specify: _____

5. Next Steps & Follow-Up

- Date and time of next appointment with adult endocrinologist: _____
- Name and contact of information of the adult endocrinologist: _____

Checklists for transition of patients with specific conditions



Identified conditions

- Congenital hypogonadotropic hypogonadism
- X-linked hypophosphatemia
- MEN1
- Prader-Willi Syndrome
- AVP deficiency
- Turner syndrome / Klinefelter syndrome
- Congenital hypothyroidism

ToC Working group e-Survey



European
Reference
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Endo-ERN



Endo-ERN 'Transition of care' survey for rare endocrine disorders

Dear Participant,

Thank you for taking the time to participate in this survey, developed by the **Endo-ERN 'Transition of Care' Working Group**.

This survey aims to explore current practices, challenges, and strategies in managing the **transition of care** for patients with **rare endocrine diseases** across different healthcare providers and institutions. The questions address key aspects of evaluation, treatment, follow-up, and multidisciplinary collaboration during the **transition from pediatric to adult care**.

Survey Details:

- Estimated completion time: **15-20 minutes**.

- All responses will be treated as **confidential** and will be used exclusively for research purposes within the framework of Endo-ERN.

At the end of the survey, you will also have the opportunity to share **comments** or **suggestions** to further enhance this research.

If you have any questions or need further clarification, please feel free to contact us at:

andrea.isidori@uniroma1.it & u.doehnert@uni-luebeck.de (Chairs)

francesco.carlomagno@uniroma1.it & matteo.spaziani@uniroma1.it (Managers)

Thank you for your valuable contribution!

Endo-ERN Transition of Care Working Group

START SURVEY NOW

1 Mapping Transition of Care for Rare Endocrine Conditions in Europe: Findings 2 from a Cross-Sectional Survey by the Endo-ERN ToC Working Group

3

4 Authors:

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12

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ToC Survey draft

- **Data analysis** (w/ C. Gaasterland) completed
- **Manuscript circulated** among ToC WG members
- **Feedback** collected and integrated
- **Submission** underway to EJE / Endo Connections



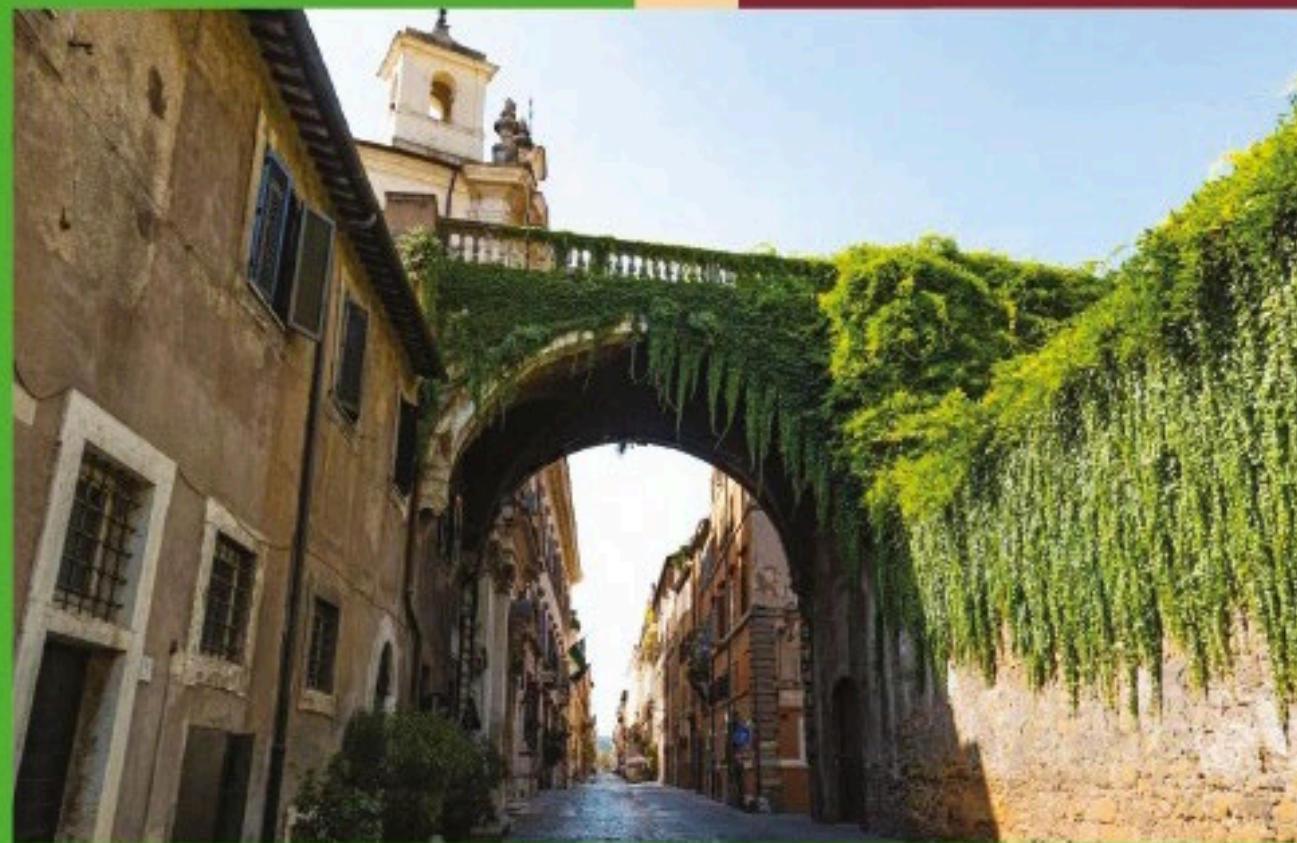
Google Drive

8th TALENT Conference Meeting



ROMA
11.12.13 FEBBRAIO
2026

Oratorio del Gonfalone



11-13th February 2026 in Rome, Italy – 3 Days full of...

- **8 Main thematic groups-specific sessions**
 - **24 lectures** with international-level speakers and **16 moderators**
- **‘Voices in Transition’**: roundtable between **physicians** and **patient representatives**

Objectives:

- 1) Highlight gaps and best practices in transition of care
- 2) Foster dialogue between clinicians and patients
- 3) Gather input for future guidelines and Endo-ERN projects

Endorsement (granted / underway) by:

- **Endo-ERN**
- European Society of Endocrinology (**ESE**)
- European Academy of Andrology (**EAA**)
- European Neuroendocrine Association (**ENEA**)
- Italian Society of Endocrinology (**SIE**), Italian Society of Diabetology (**SID**), Italian Society of Andrology and Sexual medicine (**SIAMS**), Italian Society of Paediatric Endocrinology and Diabetology (**SIEDP**), etc...

Cooperation

ERN Overarching Transition Group

- ✓ Regular Meetings (next 02/09/2025)
- ✓ Workshop in Ghent (27/02 - 01/03/2026)

Participation of Paediatric, Adult and Young
Patient Representative

- ✓ ERN Overarching Transition Group Survey

ESE-ESPE Transition of Care Working Group

- ✓ Monthly meetings
- ✓ Recommendations close to submission

Other RD Project updates

Charlotte van Beuzekom, Project Manager Endo-ERN



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Other RD project updates

JARDIN

- Task 6.2, care pathway development
- JARDIN surveys



ERDERA

- Networking Support Scheme
- General Assembly 29-31 October 2025, Amsterdam UMC



ERDERA Funding

The ERDERA Networking Support Scheme (NSS) Call **is open!**

[CLICK HERE](#)

www.erdera.org

AOB & questions



