



European  
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Endo-ERN

# Endo-ERN Steering Committee/Advisory Board Meeting

Date: October 1, 2024

Time: 9:30 –11:00 CET



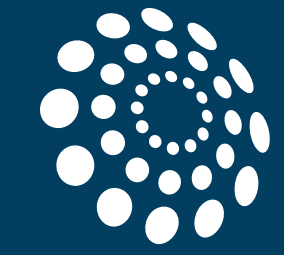
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## Agenda

09:30 – 09:35 Welcome

09:35 – 11:00 Presentation WP activities incl. Discussion

*8 minutes planned per Work package & Working Group*



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## WP1 Management & Coordination

**Alberto Pereira**, Adult Chair Endo-ERN

**Olaf Hiort**, Paediatric Chair Endo-ERN

**Charlotte van Beuzekom**, Project Manager Endo-ERN



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## Replacement (chair) positions

MTG	Current chair	New chair	Institute	Country	Starting
7	Luca Persani (adult) – IT	Claus Gravholt	Aarhus University Hospital	Denmark	01 October 2024



**Claus Gravholt**

## Collaborations

- Endocrine Connections – special issue
- EURORDIS Supporting Partner Agreement
- ESPE Rare Disease Advisory Committee (Evangelia Charmandari / Marek Niedziela)
- Visit European Commission to Amsterdam UMC
- Notebook pilot (Petra Bruegmann)

Pilot : **Notebook** - a joint project between Endo-ERN und Eurordis to evaluate and support the impact of patients representatives (epags)

# ERN Evaluation Technical Toolbox



## **1.3. The ERN has established mechanisms for the integration of patient organizations in the strategic actions.**

### **Guidelines**

It is essential to identify the different patient organizations related in one way or another to the areas of interest of the ERN, and to establish contact with as many as possible as long as they have a recognized level of representation.

The level of participation and the degree of collaboration of these patient organizations should be evaluated periodically to understand the nature of the collaboration and the need, if any, to introduce changes in the collaboration mechanisms.

# Monitor & Evaluate Patient Involvement in ERN



## Measurable Elements

**1.3.1. Patient representatives have been included in the governance framework of the ERN.** 

**Evidence** Describe the specific role of patient representatives in the current governance of the Network (where and how they are involved).



**1.3.2. The Board has incorporated the opinion of patients and families when outlining strategies.** 

**Evidence** Explain in which issues the patients and families' input has been requested and which opinions were incorporated.



**1.3.3. Patients and support groups are major stakeholders in ERN-related activities.** 

**Evidence** a) Is this participation formal and continuous? Provide some examples.  
b) Acknowledgement of patients' coauthorship in ERN deliverables



**4.2.1. The ERN collaborates with patient organisations to develop and implement care pathways, guidelines, protocols, and indicators.** 

**Evidence** List of actively involved patient organisations and activities developed with them (i.e., development of clinical guidelines, development, and implementation of clinical indicators...)  
  
Acknowledgment of co-authorship in guidelines or other documents produced.  
  
Related measurements included in the e-PAG Impact Assessment Framework, if used.





### 1.3.4. The ERN monitors and evaluates the involvement of patients in the activities of the ERN

**Evidence** Specific methodology to collect information regarding the participation of patient representatives in the ERN activities (i.e., satisfaction surveys, specific monitoring indicators).

Attach the outcomes

# Notebook structure



[Add ERN Log]



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## The impact of my involvement in the ERN [insert your name] Notebook 2024

Date DD-MM-YY	Name of Project or Activity	Contribution (for ex. contributed to initial focus of the project, contributed to project meeting discussions, help the project to focus on patient needs, contributed ideas to the project, contributed to recommendation or decisions in the project, etc.)	Impact Please share examples of how your contribution made a difference

# How the notebook can be used (my example)



Date	Name of project	Contribution	Impact
February 2024	S/A Meeting	<ul style="list-style-type: none"> <li>Slides with results of EMENAs MEN survey</li> </ul>	<ul style="list-style-type: none"> <li>Patients' views on unmet needs shared to inform discussion.</li> </ul>
May 2024	General Assembly, Milan	<ul style="list-style-type: none"> <li>Contribution to the MTG 4 Session &amp; Presentation</li> </ul>	<ul style="list-style-type: none"> <li>Patients' views on guidelines and registries.</li> </ul>
May 2024	EuRRECa	<ul style="list-style-type: none"> <li>Contribution to symposium, poster and presentation</li> <li>Insisting on MEN 1 module</li> </ul>	<ul style="list-style-type: none"> <li>Presenting patient views' view on registries as results of the MEN survey.</li> <li>It is in discussion and will be delivered.</li> </ul>
23 May 2024	Pathways	<ul style="list-style-type: none"> <li>Start of the discussion on MEN 1 Patients' Journey &amp; Pathway.</li> </ul>	<ul style="list-style-type: none"> <li>Insisting on the topic</li> </ul>
01 May 2024	ESE/ESPE/EndoERN/ePAG	<ul style="list-style-type: none"> <li>Survey on drugs and medicines shortages, three different drafts for the different groups (ESC national societies, ESPE national societies, European PO with endocrine conditions).</li> </ul>	Discussion with ESE and Johan de Graaf – compiling the drafts into an EU survey tool.



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## Transversal Working Group on Genomics

**Thomas Eggermann** (University Hospital Aachen, Germany)

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



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## Transversal WG „Genomic Testing“

Thomas Eggermann, (MTG 5)  
Institute for Human Genetics  
and Genome Medicine, RWTH  
University Aachen, Germany

Dirk Prawitt, (MTG 8)  
Center for Paediatrics and  
Adolescent Medicine,  
University Medical Center,  
Mainz, Germany

## What has happened so far?

- ESHG has contacted the ENDO-ERN to orchestrate the genetic issues in the different ERNs, the virtual meeting will be held next week
- Information about the Transversal group is on the ENDO-ERN website

### Genetic Testing



Led by **Thomas Eggermann**, Institute for Human Genetics and Genome Medicine, RWTH University Aachen, Germany and **Dirk Prawitt**, Center for Paediatrics and Adolescent Medicine, University Medical Center, Mainz, Germany, the initial activities are getting started. If you are interested in being involved please contact the [Endo-ERN project office](#).

The transversal working group will meet online every three months.

Purpose/aims of the working group:

- To **improve guidelines**, identify gaps and update where necessary
- Smooth and **applicable genetic testing guidelines**, which consider daily routine and the best standard for genetic testing ensuring cooperation with other societies and avoiding double work
- Suggest a **standardised genetic testing regime** whenever possible to guarantee a standardised testing procedure all over Europe
- To orchestrate and to channel the activities of Endo-ERN in the field of genetics, to **avoid fragmentation and unhelpful developments in genetic testing**

Check out the [presentation](#) from the Endo-ERN GA for an overview of planned activities. Find out additional information about the origin of the group and a continuously updated list of topics.

## 1st outcomes from the 1<sup>st</sup> virtual meeting

- Major topics which will be discussed with the TWG members in Oct. 2024:
  - „position paper“ consented with ESHG, about:
    - Standardised genetic testing procedures
    - How to achieve a standardized genetic testing quality?
    - Standardized reports addressing the needs of the clinicians
    - Germline and somatic genetic testing has to be considered
    - How to deal with VUS, incidental findings....
    - Recall of VUS
    - Common research activities
  - Overview (Matrix?) about the genetic tests (diagnostics, research) offered by ENDO-ERN members (posted in the CPMS area?)
  - Molecular board within the ENDO-ERN

## Feel free to contact us at any time!

- Ideas/Suggestions/Contacts are welcome, preferably from every MTG
- Contact:  
Thomas Eggermann  
([teggermann@ukaachen.de](mailto:teggermann@ukaachen.de))  
Dirk Prawitt  
([dprawitt@uni-mainz.de](mailto:dprawitt@uni-mainz.de))



# Transversal Working Group on Transition in Care

**Andrea Isidori** (Azienda Ospedaliero-Universitaria Policlinico Umberto I)

## Transition of Care Working Group: activities

### Completed:

- MD Participant Selection: Involvement from all MTGs, plus expressions of interest, wide representation
- Formal Approval and Kick-off Meeting (Large Attendance): Held on 24.09.2024
- Development of Visual Identity
- Shared Drive Creation

### In Progress:

- EPAGs Involvement

### Approved Tasks Underway:

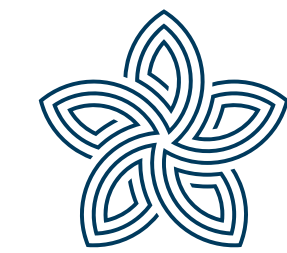
- 1.Collection of all relevant materials (papers, consensus documents, etc.) related to ToC in Rare Diseases
- 2.Development of an eSurvey to map ToC across EU countries, HCPs and MTGs
- 3.Drafting of the “First International Consultation on ToC for Rare Endocrine Disorders”: consensus on best practices and policies for ToC in ERN setting the common standards, the quality check, and the steps to improve ToC across EU.

**4.Save the Date:** TALENT Meeting scheduled for 12-14 February 2025 in Rome, Italy

# 1. Composition of the Working Group



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N	MTG / Role	Representative	Ped/Adu	E-mail (transitionwg@endo-ern.eu)
1	Coordinator	Andrea Isidori	Adu	<a href="mailto:andrea.isidori@uniroma1.it">andrea.isidori@uniroma1.it</a>
2	Coordinator	Ulla Döhnert	Ped	<a href="mailto:u.doehnert@uni-luebeck.de">u.doehnert@uni-luebeck.de</a>
3	1/Adrenal	Svetlana Lajic	Ped	<a href="mailto:svetlana.lajic@ki.se">svetlana.lajic@ki.se</a>
4	1/Adrenal	Nicole Reisch	Adu	<a href="mailto:Nicole.Reisch@med.uni-muenchen.de">Nicole.Reisch@med.uni-muenchen.de</a>
5	1/Adrenal	Uta Neumann	Ped	<a href="mailto:uta.neumann@charite.de">uta.neumann@charite.de</a>
6	2/Calcium&Phosphate	Corinna Grasemann	Ped	<a href="mailto:corinna.grasemann@rub.de">corinna.grasemann@rub.de</a>
7	2/Calcium&Phosphate	Eva Kassi	Adu	<a href="mailto:ekassi@med.uoa.gr">ekassi@med.uoa.gr</a>
8	2/Calcium&Phosphate	Graziamaria Ubertini	Ped	<a href="mailto:graziamaria.ubertini@opbg.net">graziamaria.ubertini@opbg.net</a>
9	3/Glucose&Insulin	Felix Reschke	Ped	<a href="mailto:felix.reschke@hka.de">felix.reschke@hka.de</a>
10	4/Genetic tumours	Annemarie Verrijn-Stuart	-	<a href="mailto:a.a.verrijnstuart@umcutrecht.nl">a.a.verrijnstuart@umcutrecht.nl</a>
11	5/Growth&Obesity	Susan O' Connell	Ped	<a href="mailto:susan.oconnell@childrenshealthireland.ie">susan.oconnell@childrenshealthireland.ie</a>
12	5/Growth&Obesity	Claudia Giavoli	Mix	<a href="mailto:claudia.giavoli@unimi.it">claudia.giavoli@unimi.it</a>
13	5/Growth&Obesity	Danilo Fintini	Ped	<a href="mailto:danilo.fintini@opbg.net">danilo.fintini@opbg.net</a>
14	6/Hypothal&Pit	Evangelia Charmandari	Ped	<a href="mailto:evangelia.charmandari@googlemail.com">evangelia.charmandari@googlemail.com</a>
15	6/Hypothal&Pit	Nienke Biermasz	Adu	<a href="mailto:n.r.biermasz@lumc.nl">n.r.biermasz@lumc.nl</a>
16	6/Hypothal&Pit	Violeta Iotova	Mix	<a href="mailto:iotova_v@yahoo.com">iotova_v@yahoo.com</a> , <a href="mailto:violeta.iotova@mu-varna.bg">violeta.iotova@mu-varna.bg</a>
17	7/DSD	Hedi Claahsen	Ped	<a href="mailto:Hedi.Claahsen@radboudumc.nl">Hedi.Claahsen@radboudumc.nl</a>
18	7/DSD	Claus Gravholt	Mix	<a href="mailto:claus.gravholt@clin.au.dk">claus.gravholt@clin.au.dk</a>
19	8/Thyroid	Dana Craiu	-	<a href="mailto:dcraiu@yahoo.com">dcraiu@yahoo.com</a>
20	8/Thyroid	Andreea Badea	-	<a href="mailto:dr.andreea.badea@gmail.com">dr.andreea.badea@gmail.com</a>
21	Manager	Francesco Carlomagno	Mix	<a href="mailto:francesco.carlomagno@uniroma1.it">francesco.carlomagno@uniroma1.it</a>
22	Manager	Matteo Spaziani	Mix	<a href="mailto:matteo.spaziani@uniroma1.it">matteo.spaziani@uniroma1.it</a>

**To be  
implemented  
with EPAGs**

## 2. Visual Identity



Endo-ERN

**TALENT**

TRANSITION OF CARE  
WORKING GROUP



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**TALENT**

TRANSITION OF CARE  
WORKING GROUP

### 3. Transition of care Working group: *Toward Synergy*

Implement the available resources, and collaborate with

- Other ERNs

And

All reference scientific societies to merge efforts:

- ESE: Clinical committee & Rare disease committee
- ESPE: Clinical practice committee & Rare disease advisory committee
- ISPAD (rare forms of diabetes)?
- Endocrine society?





# WP2 Communication & Dissemination

Aimee Casey, Communication Manager, Endo-ERN

Endo-ERN is a network connecting patients and healthcare providers across Europe, aiming to provide knowledge and resources for diagnosis and treatment of rare endocrine conditions.

Read More

Watch video



### Endo-ERN News

European Society of Endocrinology Endo-ERN

**PARTICIPATE: NEW ESE POSITION STATEMENT Adrenal Insufficiency (AI)**

Attention Endo-ERN representatives! You are invited to participate in this important two-year project to develop a new ESE position statement:

[READ MORE](#)



**APPOINTED: Johan de Graaf appointed to EURORDIS Board**

Great news! Endo-ERN ePAG Johan de Graaf has been elected to EURORDIS Board of Directors during the organisation's annual General

[READ MORE](#)



**WELCOME: NEW ePAG Esther**



## Social media update since GA

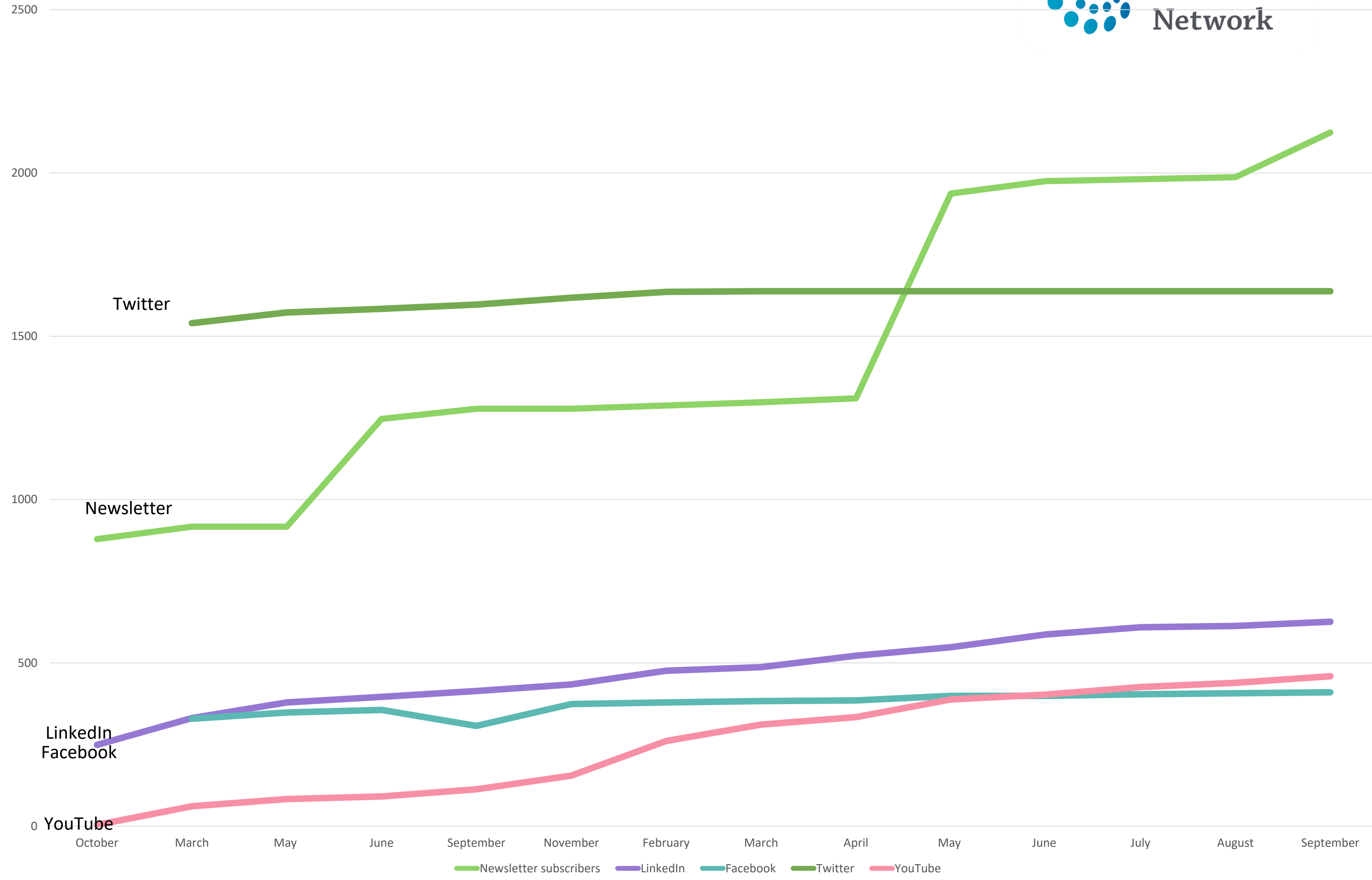


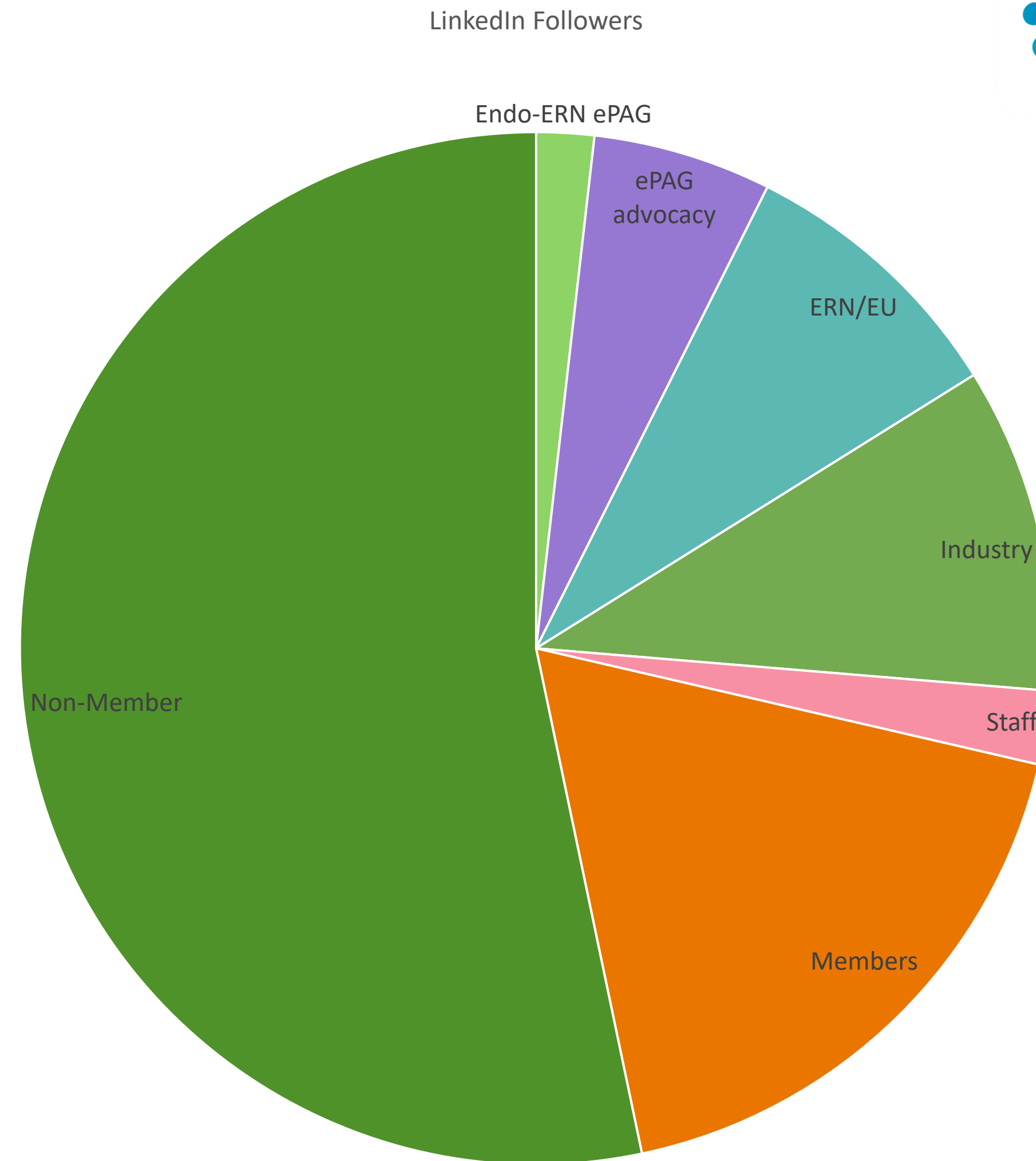
	Total	Increase
LinkedIn	626	+104
Facebook	410	+25
YouTube	459	+125
Newsletter subs	2124	+814



# Social Media

User development





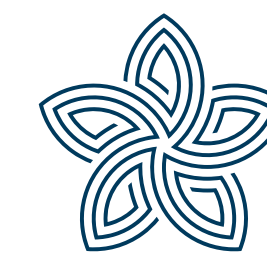
■ Endo-ERN ePAG ■ ePAG advocacy ■ ERN/EU ■ Industry ■ Staff ■ Members ■ Non-Member



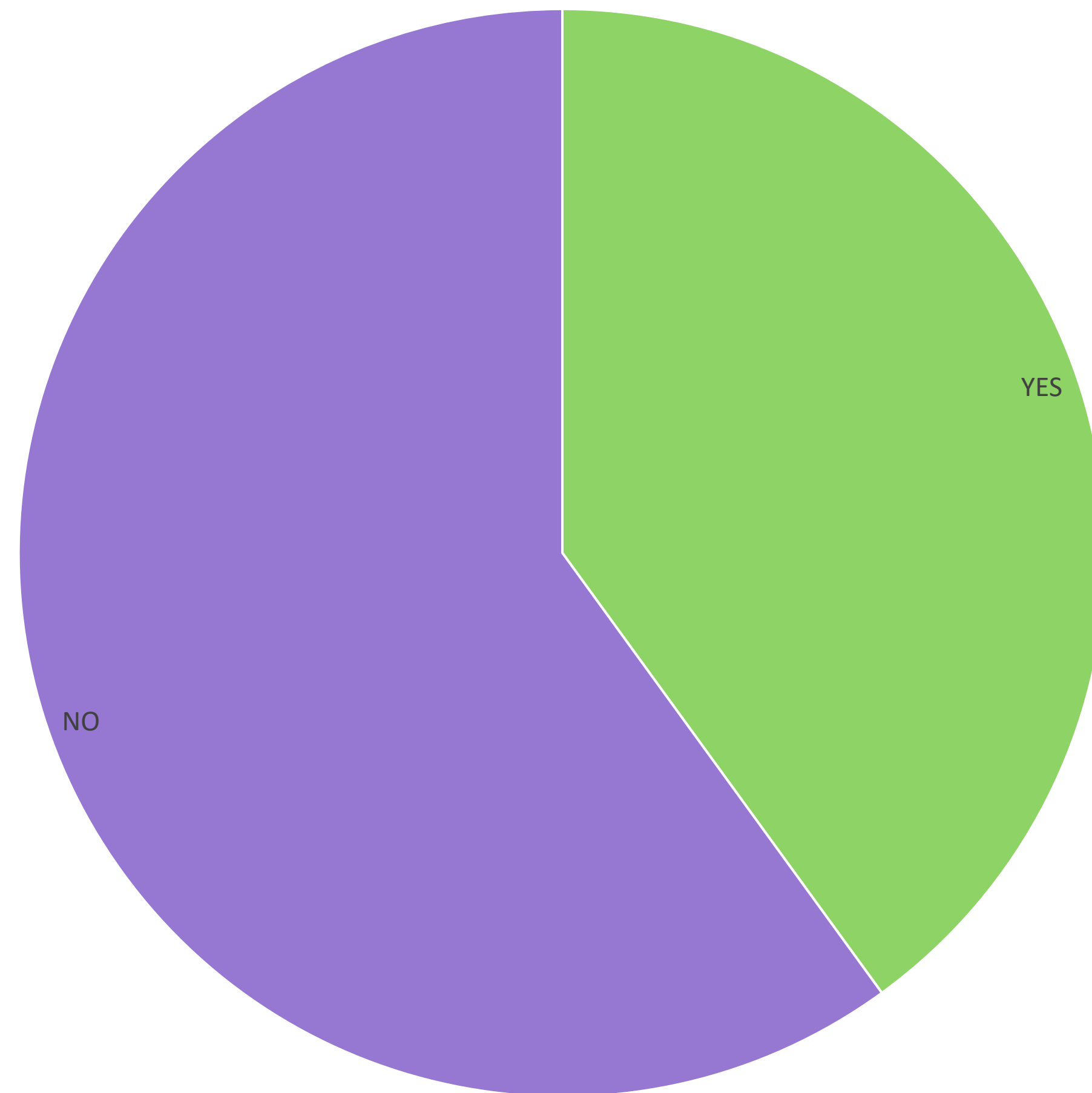
SC/AB Members on LinkedIn



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■ YES ■ NO



# Endo-ERN Publications Database



## Filter list of publications

<b>Condition / Disease</b>	<input type="text"/>	<b>Author(s)</b>	<input type="text"/>
<b>MTG</b>	Select option ▼	<b>Keyword</b>	<input type="text"/>
<b>Year</b>	Select option ▼	<b>ORPHAcode / other</b>	<input type="text"/>
<b>Affiliated Institution / ERN</b>			
Select option ▼		<b>Apply filter</b>	<b>Reset</b>

481 publication(s) found matching the search criteria

Sort results by ▼

## Primary Hypothyroidism in Childhood Cancer Survivors Treated With Radiation Therapy: A PENTEC Comprehensive Review

**AUTHOR(S):** Milano MT, Vargo JA, Yorke ED, Ronckers CM, Kremer LC, Chafe SM, van Santen HM, Marks LB, Bentzen SM, Constine LS, Vogelius IR

**AFFILIATED INSTITUTION / ERN:** Amsterdam UMC ; University Medical Center Utrecht ; Copenhagen University Hospital, Rigshospitalet ;

**CONDITION / DISEASE:** Pediatric primary hypothyroidism

**PUBLICATION:** International Journal of Radiation Oncology\*Biology\*Physics v119.2 p482-493 **YEAR:** 2024 **ORPHACODE /**

**OTHER:**

**DOI:** [10.1016/j.ijrobp.2021.02.001](https://doi.org/10.1016/j.ijrobp.2021.02.001) **KEYWORDS:**



- Publication database is now live with data until end of 2021



# Rare Disease Day 2025

Friday, 28 February

[Endo-ERN Rare Disease Day Poster - Endo-ERN](#)

**RARE DISEASE DAY** **1 IN 2000 PEOPLE**  
**28 FEBRUARY 2023** **LIVE WITH A RARE DISEASE**

Did you know that **rare diseases** affect the daily lives of up to **36 million** people in the EU?

**This hospital** is a member of **Endo-ERN**, a virtual network of **111 Reference Centres (RCs)** in **28 countries\*** that offer access to clinical experts for patients with **rare endocrine conditions**.

The objective of **Endo-ERN** and all European Reference Networks (ERNs) is to ensure that it is the **medical knowledge and expertise that travel, rather than the patients**.

**Secure Virtual Consultations**  
with international experts, no travel required using the Clinical Patient Management System (CPMS).

**Rare Disease Registries**  
offer access to high-quality, patient-centered registries for rare endocrine conditions. The European Registries For Rare Endocrine Conditions are called EURRECs.

**European Patient Advocacy Groups (ePAGs)**  
represent patients within Endo-ERN. The ePAGs help communicate the availability of virtual consultations, rare disease registries and helpful patient information to their members.

**Rare Disease Research**  
including an overview of studies by our members and a listing of associated publications.

**Training & Education**  
Involves knowledge sharing between our members in-person (Exchange Programme) and on-line (webinars).

[www.endo-ern.eu](http://www.endo-ern.eu)

Do you want to know more about Endo-ERN? Sign up for our newsletter!

\*Total includes 14 affiliated partners



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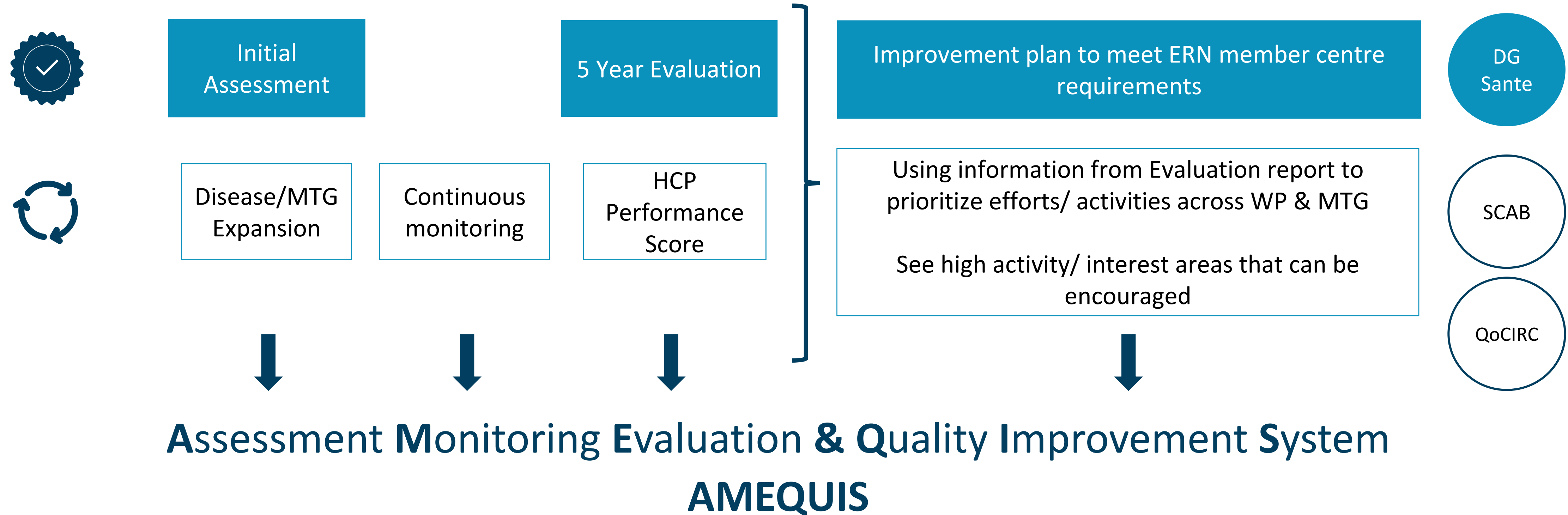
## WP3 Evaluation

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



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# Evaluation in Endo-ERN – QI



## Continuous Monitoring Indicators 2024/25

- HCPs will be also asked about HCP data to complete the HCP performance Score
- The Endo-ERN survey will have to gather input for the new grant KPIs
- **WP chairs will have chance to add Qs to this survey**

	Objective 1: To ensure that ERNs are operational and successfully carry out their activities	Source
1.1	Number of Member States represented in the ERN	EC
1.2	Number of full members of the ERN	EC
1.3	Number of affiliated partners of the ERN	EC
1.4	Number of patient organisations represented in the ERN	ERN
	Objective 2: To improve access to clinical advice, diagnosis, treatment, and follow-up of patients	
2.1	Number of new patients referred to the Health Care Providers of the ERN, with the diagnosis of a disease or condition that falls within the scope of the ERN	ERN
2.2	Number of patient cases discussed in the CPMS	EC
	Objective 3: To optimise patient outcomes by combining healthcare professionals' skills & resources used	
3.1	Number of CPMS outcome reports produced during the reporting period	EC
3.2	Days between enrolment in the CPMS and multidisciplinary clinical advice 3.2.a) non-urgent cases 3.2.b) urgent cases	EC
	Objective 4: To increase the capacity of professionals to recognize and manage cases of rare and complex conditions and diseases within the scope of the ERN	
4.1	Number of education/training activities not accruing higher education credits aimed at healthcare professionals delivered by the coordination teams or HCP members of the ERN	ERN
4.2	Number of formal educational activities that are accruing higher educational credits aimed at healthcare professionals delivered by the coordination teams or HCP members of the ERN	ERN
	Objective 5: To reinforce clinical research in the field of rare and complex conditions and diseases by collecting data and carrying out research activities	
5.1	5.1.a) Number of Clinical Trials involving ERN members in at least two Member States 5.1.b) Number of Observational prospective studies / Observational cohort or case-control studies / Case-series studies involving ERN members in at least two Member States	ERN
5.2	Number of accepted peer-reviewed publications in accredited scientific journals falling within the scope of the ERN and acknowledging the ERN.	ERN
	Objective 6: To ensure that patients referred to ERNs have equal access to high and quality healthcare services	
6.1	Number of Clinical Practice Guidelines and other types of Clinical Decision-making Tools, adopted for diseases within the scope of the ERN	ERN
6.2.	6.2.a) Number of new Clinical Practice Guidelines written by the ERN and released during the reporting period 6.2.b) Number of other types of new Clinical Decision-making Tools (clinical consensus statements or consensus recommendations), written by the ERN and released during the reporting period	ERN
	Objective 7: To guarantee that knowledge, expertise and awareness is spread outside the ERN so that more people can benefit from the ERN activities	
7.1	Number of congresses/conferences/meetings at which the ERN activities and results were presented	ERN
7.2	Number of individual ERN website hits	ERN
	Objective 8: Complex and long-term indicators which need further development	
8.1	Level of patient satisfaction (still to be developed)	
8.2	Integration in the national health systems of Member States (still to be developed)	



2023 = 18019 via EC platform

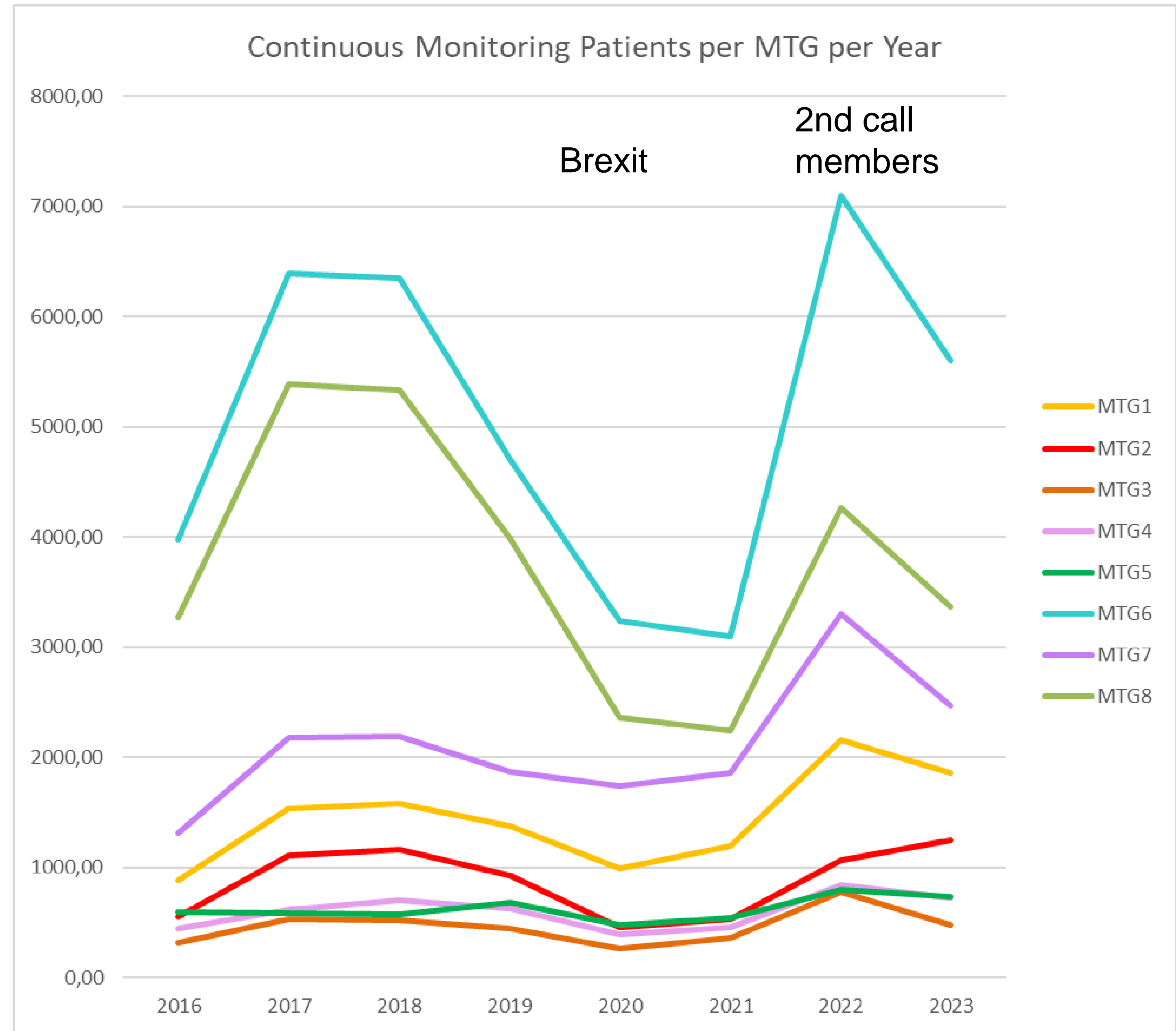
- 16479 e-REC in 2023

**Missing patients = 1540 from 32 HCPs**

- (More HCPs have e-REC users now)

**Key Reminder/Focus for members:**

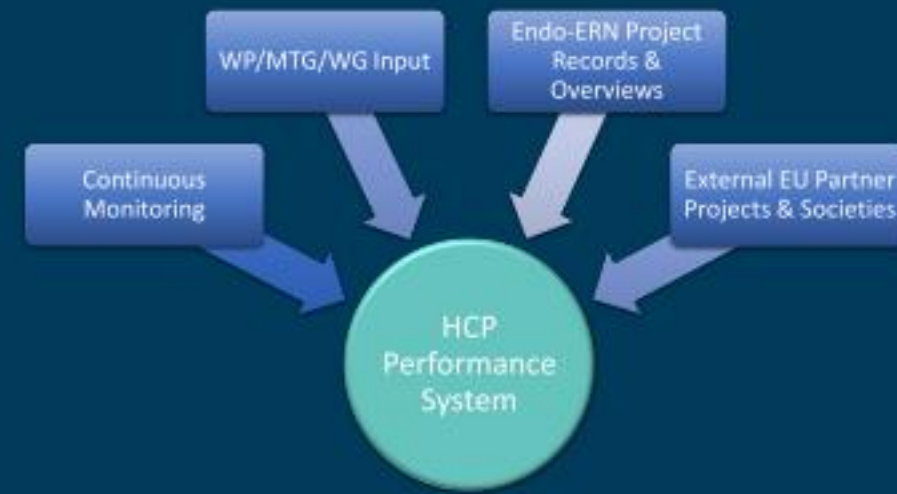
- **Update e-REC before end of year**





## Endo-ERN HCP Performance Report 2023

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.



From our Continuous Monitoring exercise, in 2023 our network members have...

### 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

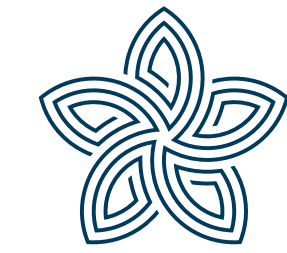
21 pts **mandatory** = 60% of total score  
 20 pts **expected** = 25% of total score  
 22 pts **bonus** = 15% of total score

The resulting weighted values are used to calculate the overall Overall Score. Please see [our website](#) for further details.

- 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)



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## HCP Performance Reports

All HCPs in Endo-ERN received their reports.

### General Feedback:

- 10 HCPs received updated reports/ scoring
- 2 updates pending

Excellent = 56 (54%)

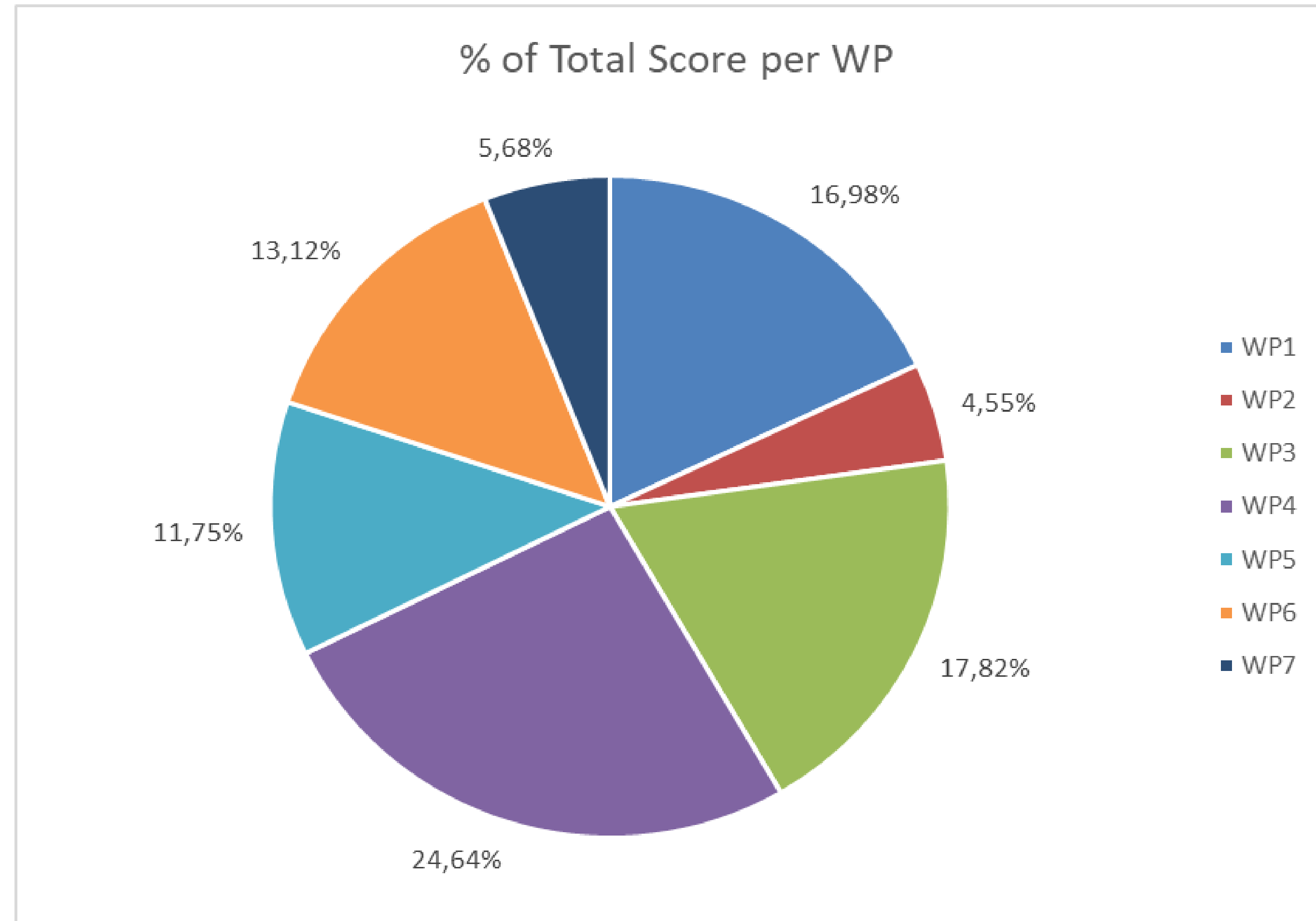
Good = 46 (44%)

Poor = 3 (>3%)

### Positive reaction to the scoring and receiving feedback!

- Each **WP** will receive personal feedback in a general report
- Report will include **MTG** feedback from Continuous Monitoring

# WP Specific Breakdown HCP PS Score



# Continuous Monitoring Overview

## 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

*New feature 2025*

### How

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

*Awaiting confirmation*

### When

- Annually – once a year in the first quarter
- 2023 data collected starting from January 2024

### Outcomes

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

*Updated for 2024*



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## WP4 Healthcare & CPMS

**Olaf Hiort**, Paediatric Chair Endo-ERN

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

**Greta Ginski**, CPMS Operational Helpdesk Manager



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## New CPMS OH

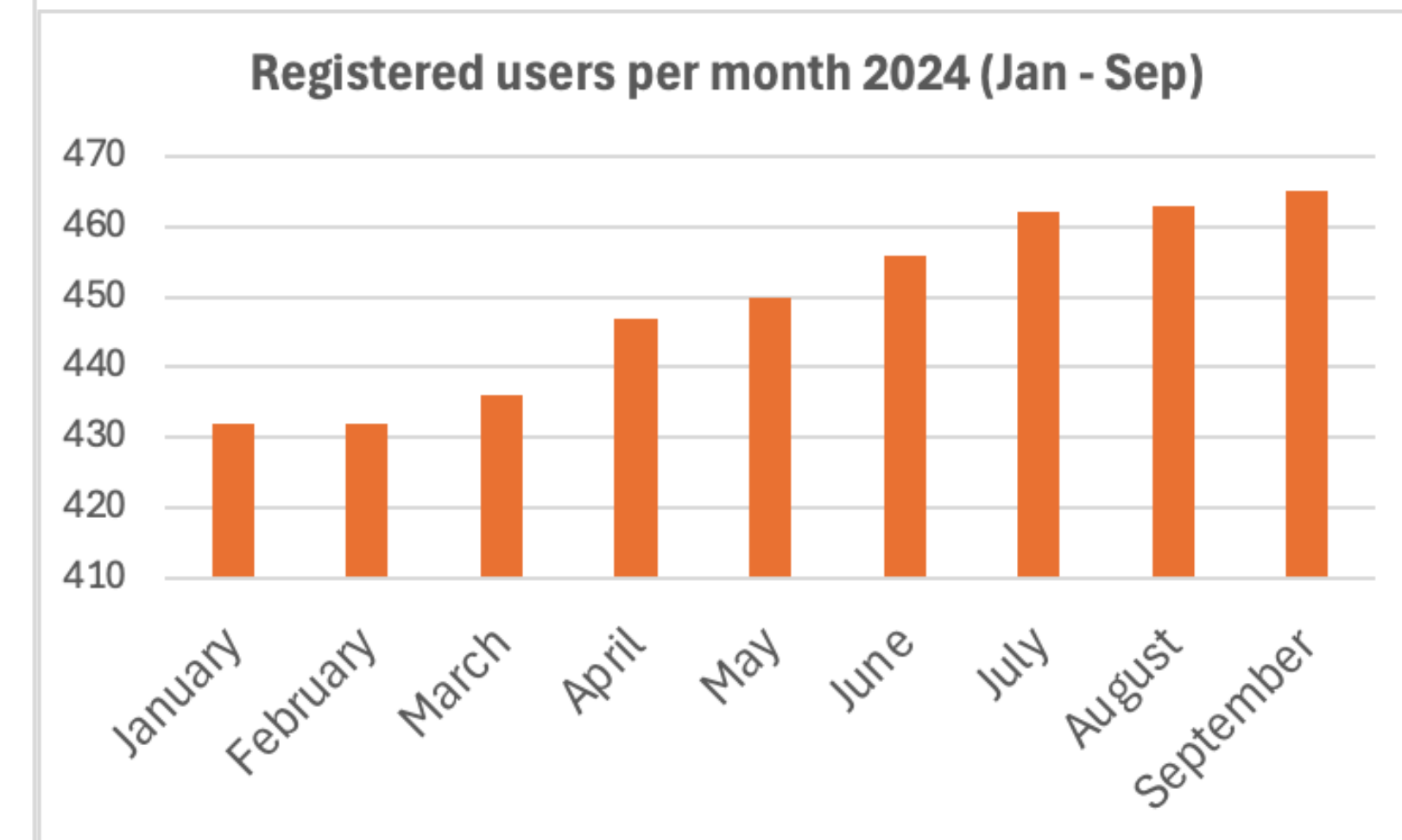
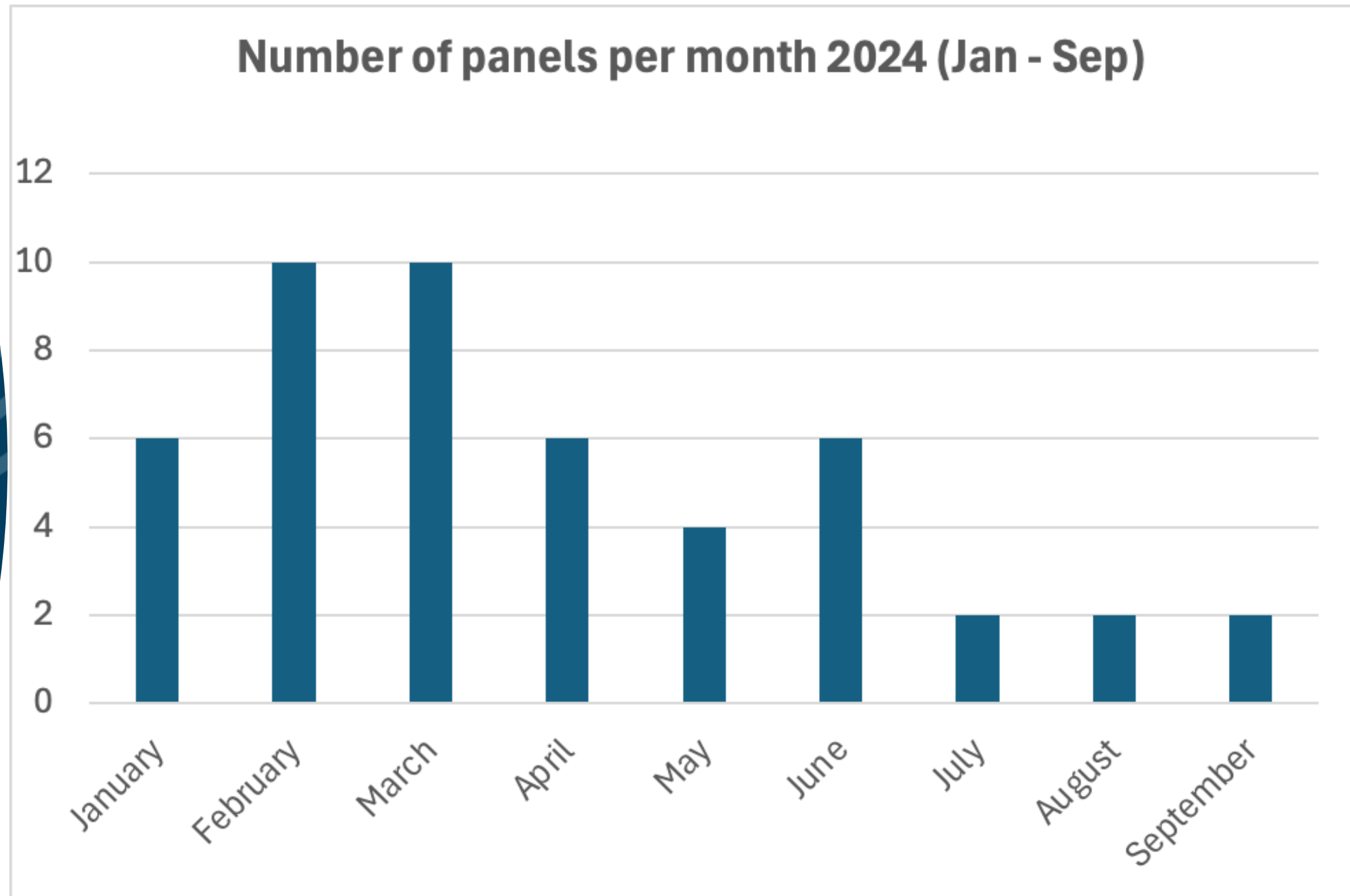
- UKSH Lübeck, Klinik für Kinder- und Jugendmedizin
- Biomedical (M.Sc.)/Health sciences (B.Sc.)



**Greta Ginski**

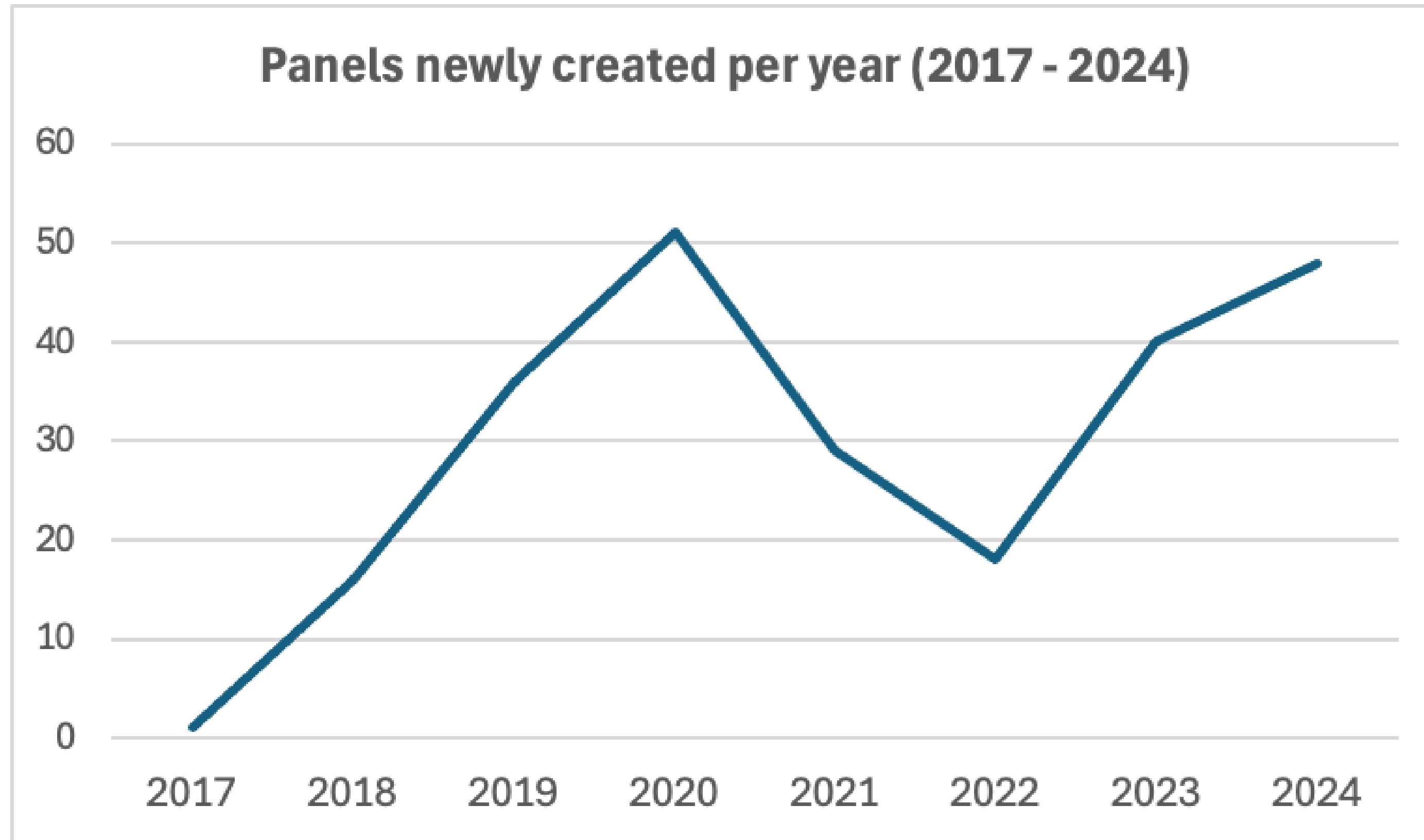
*greta.ginski@uksh.de*

# CPMS Current status



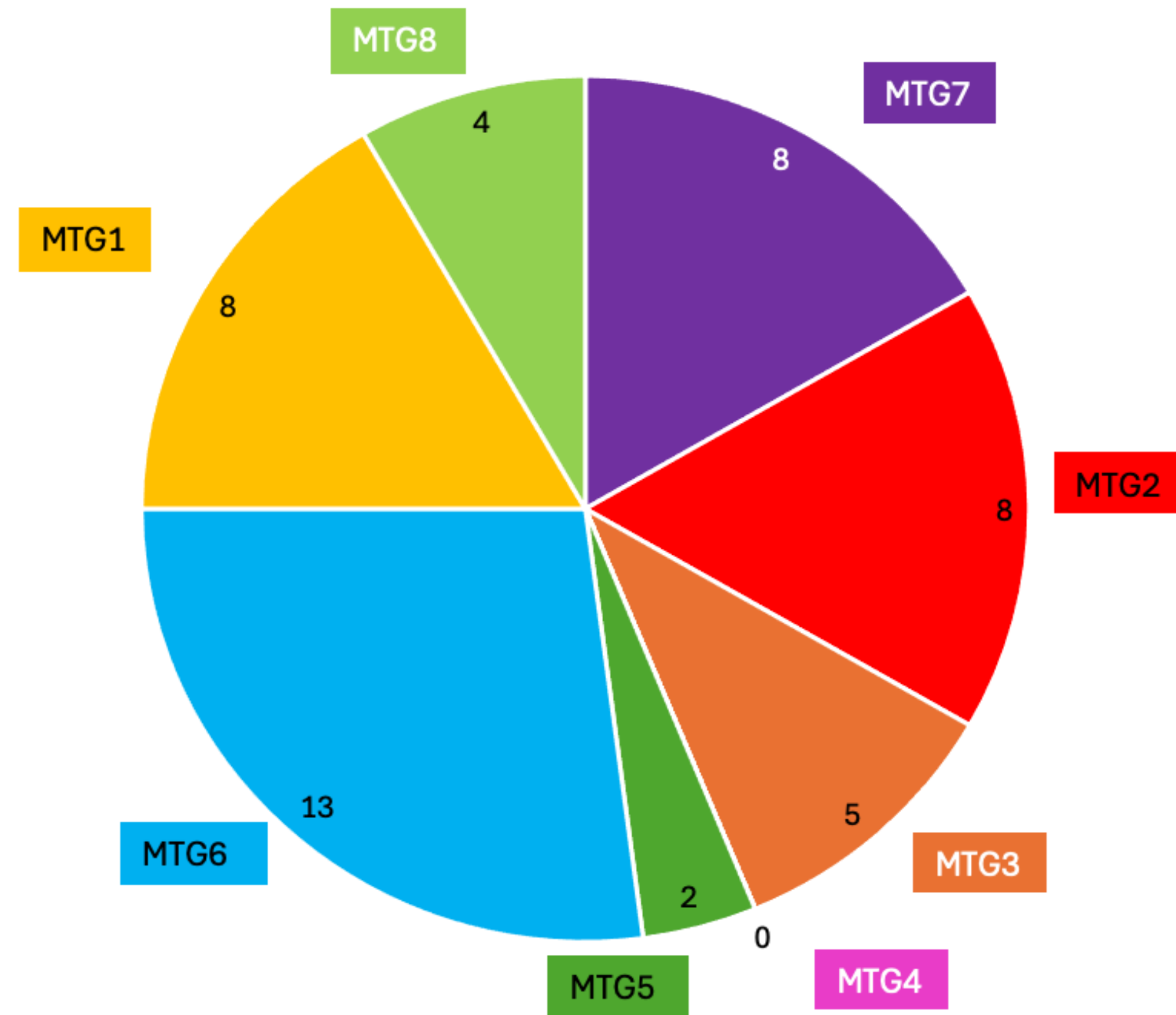
- Newly created panels in 2024: 48
- Continuous monitoring: Patients in Endo-ERN = 18.019

# CPMS Current status



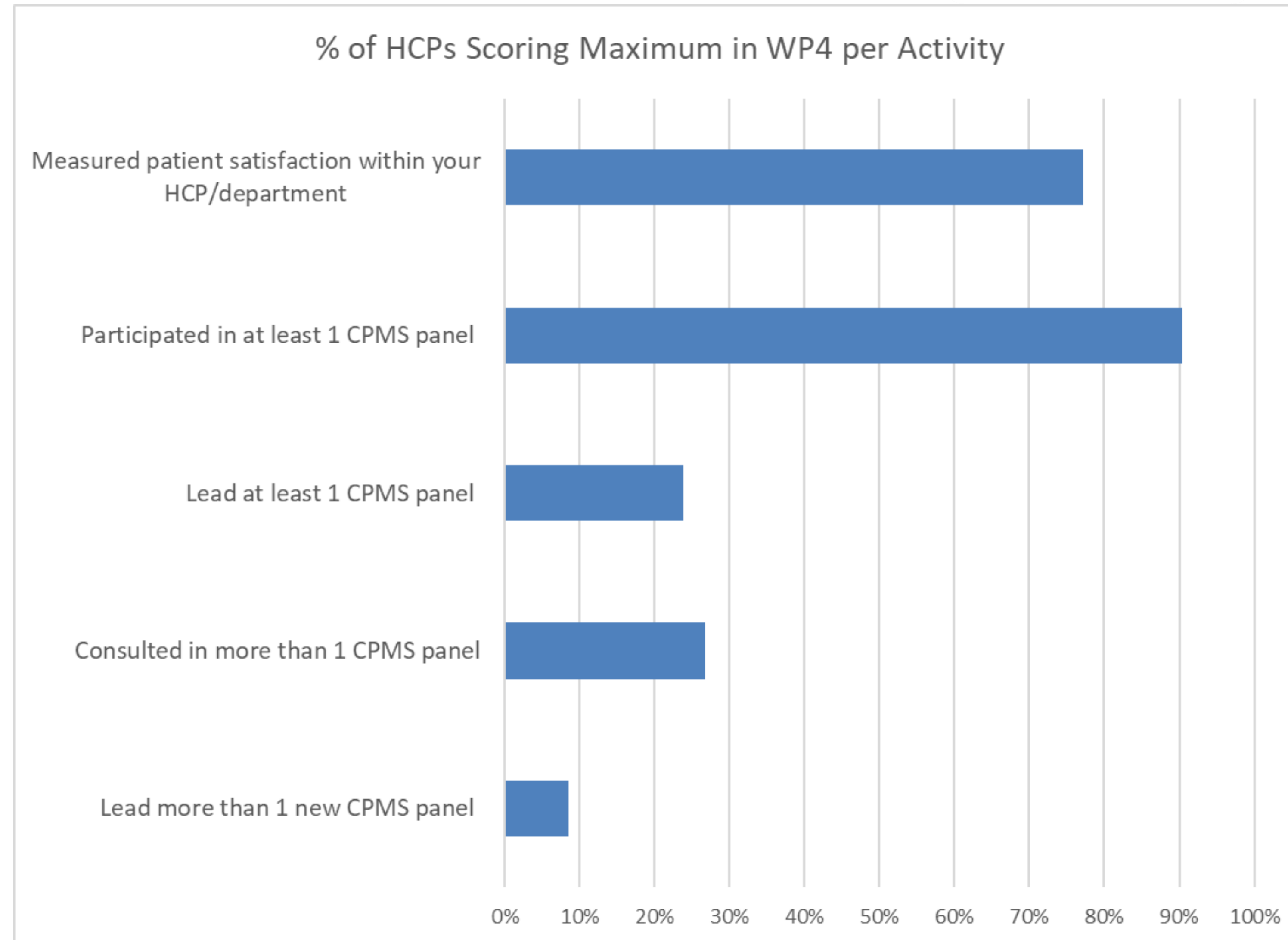
## CPMS Current status

Panels created in 2024 per MTG (Jan - Sep)



- No panels this year: **MTG4**
- MTGs without regular meeting coordination between Chairs and Helpdesk = **MTG3**, **MTG4**, **MTG5**

# WP4 HCP Performance System Scoring



## HCPs with no CPMS users

Plan of action =  
Targeted approach  
for CPMS 2.0



	HCPs with No CPMS Users
RO07	County Emergency Clinical Hospital
FR49	Reference center for rare diseases of calcium and phosphate- HEGP
HR05	Sestre milosrdnice University Hospital Center
HR07	MERKUR UNIVERSITY HOSPITAL
IT02	AOU University of Campania Luigi Vanvitelli, Naples
IT107/ IT116	Azienda Unità Sanitaria Locale-IRCCS of Reggio Emilia
IT14	AOU - Modena
IT50	IRCCS Institute Giannina Gaslini - Genoa
LV02	Pauls Stradins Clinical University Hospital
SK02	University Teaching Hospital in Martin

# CPMS 2.0



As Endo-ERN we have attended all BIG meetings to date.

- 8 ERNs will migrate first (users only) : BOND, CRANIO, GENTURIS, EpiCARE, EURO-NMD, eUROGEN, RITA, VASCERN

Our likely migration month – December 2024

- Slightly later due to custom datasets
- Current CPMS stopped by end of December
- Available to test these now in CPMS 2.0 test platform [CPMS 2.0 \(ern-net.eu\)](https://ern-net.eu)

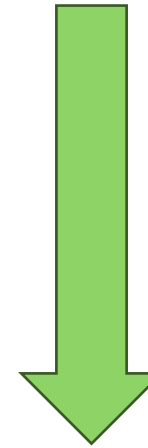
2 key preparation steps:



# CPMS 2.0 Preparations

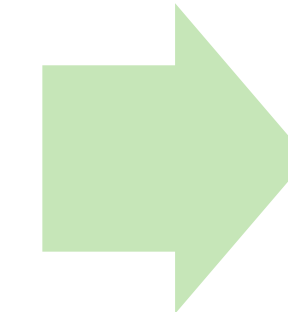


Creates all necessary documents for a hospital DPO to approve CPMS/ create DPIA is needed



## September

- Endo-ERN - DPO trial run
- OH prepares documents & Email to issue to all HCPs



## October

- HCP reps receive, then forward email to DPO
- Follow up for progress
- Ask DG Sante for support if necessary
- Agree on local consent form

- Home
- Patient Record
- Add New Patient
- Meetings
- Supporting Docu...

Search for a record > Addison's disease 01

# Addison's disease 01

P0000186 Under Discussion

- Details
- Files
- Participants
- History

## Patient Details

Sex: Female      Age: 13 Years - 10 Months

Record created  
15 February 2024

Thematic and Sub-thematic area

Adrenal

Extended details

## Healthcare provider details

ERN  
Endo-ERN

Healthcare provider  
Amsterdam University Medical Centers Location AMC

Lead  
Emily Zola (inactive)

## Discussion topic

testing on 22nd Feb

View more

Questions about patient/  
consultation reason is written  
in discussion topic.

Close discussion

- EZ** Dr. Emily Zola (inactive) February 22, 2024 - 10:06  
Hello
- RL** Rachel Law (inactive) (on behalf of Stephane Andreasen (inactive) ) February 23, 2024 - 13:59  
HELLO back
- PD** Pascale De Keyzer (inactive) (on behalf of Elise Vanormelingen (inactive) ) February 23, 2024 - 14:02  
Come on tester

Rich text editor toolbar with icons for Bold, Italic, Underline, Strikethrough, Text color, Background color, Bulleted list, Numbered list, Indent, Outdent, Link, Unlink, Undo, Redo.

Type a new message @ to mention someone

Press Enter to send a new message

# Patient Journeys Update

Stages of Journey	Core elements
Pre- diagnosis / Onset of Symptoms Diagnosis First intervention Surveillance Treatment and/or surgery	Timeframe Clinical presentation Needs Ideal support & best practice
<b>Transition from childhood to adolescence            /adulthood/older adult            Follow up care e-natal or newborn screening</b>	<b>Wellbeing – How it felt            - Quality of Life            Significant changes from previous stage</b>

Focus for end of year:

- Acromegaly (update)
- MEN1 (new)

## WP4 - Key for MTG meetings



Need our MTG chairs to lead the way!

### 1. Close open panels

- Log into your account at least once before end of 2024 to make sure you have ACTIVE status

### 2. Start process to get **CPMS 2.0 use permission** in your HCP

### 3. **MTG regular meetings** before the end of 2024 listed here

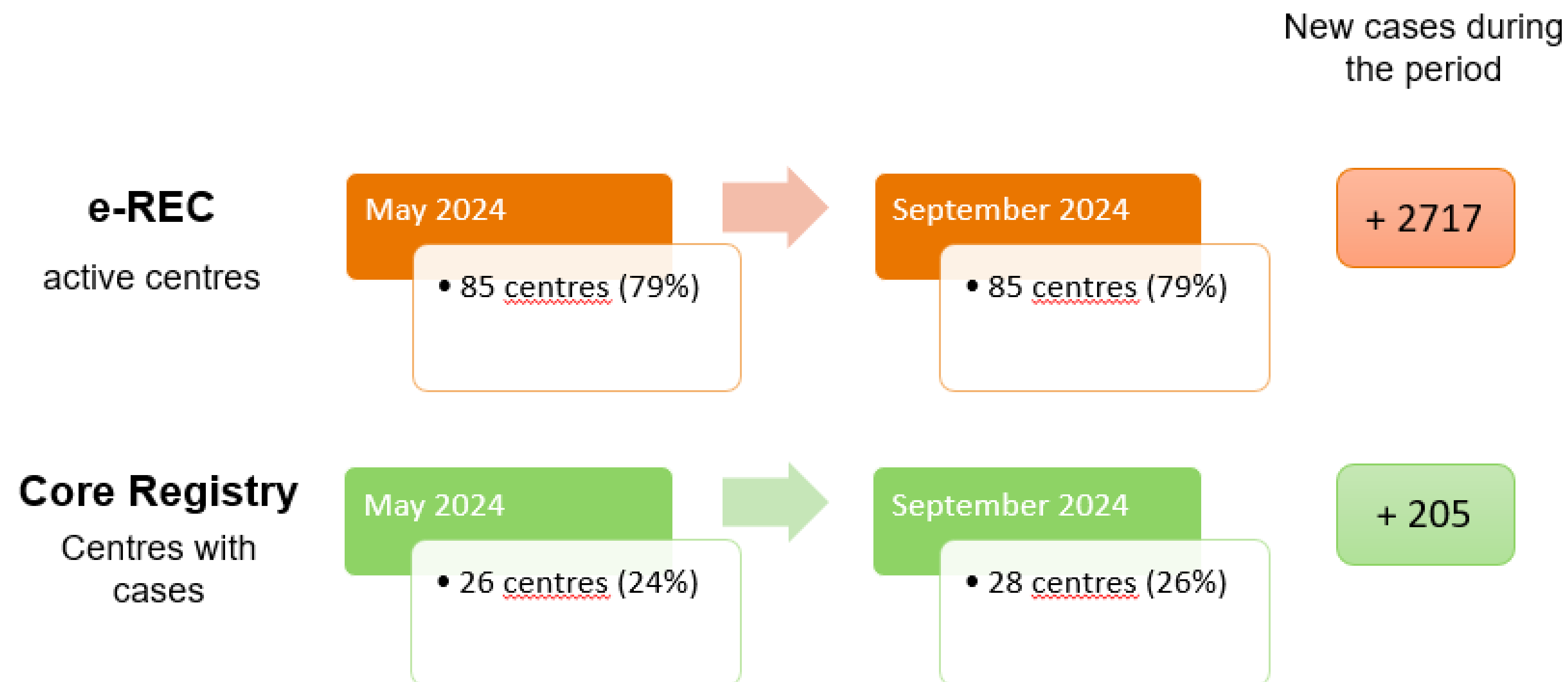
- To reserve a timeslot contact [cpmshelpdesk@endo.ern-net.eu](mailto:cpmshelpdesk@endo.ern-net.eu)
- No specific patient case? Can use time for care related discussion e.g. MTG7 discussed *DSD and genital surgery*

4. View **CPMS 2.0 custom dataset** in acceptance system/ in screenshots and offer feedback if you want changes made [CPMS 2.0 \(ern-net.eu\)](https://ern-net.eu)

## WP5 Registries, data management and analysis

Natasha Appelman-Dijkstra, Faisal Ahmed, Mariya Cherenko, Chris Breen,  
Giorgio Dal Maso, Johan de Graaf, Martha Kirchoff, Arlene Smyth

## Endo-ERN members activity



## Quality Committee Activities

- Virtual Platform onboarding

We will continue to work towards level 2 of engagement → queries to the e-REC database: diagnosis (Orphacode), number of new cases, <18 y, ≥18 y

- Security Impact Assessment of the EuRREB registries

Conducted within the LUMC to ensure compliance with international (ISO27001) and local security standards

## Key Performance Indicators (KPIs)

Implement in the Core Registry at two levels:

- General: applicable to all the conditions covered and collected as part of the Common Data Elements.  
e.g. Do you assess quality of life in this patient?
- Within Condition-specific module: developed together with working and study groups within EuRREB.



## **Affiliated registries and continuous monitoring**

- Affiliated registries listed on [eurreb.eu](http://eurreb.eu)  
(4 at the moment – Eclip, Globe-Reg, I-DSD, Crescnet)
- Online Application Form for Affiliate Status
- No need for double entries



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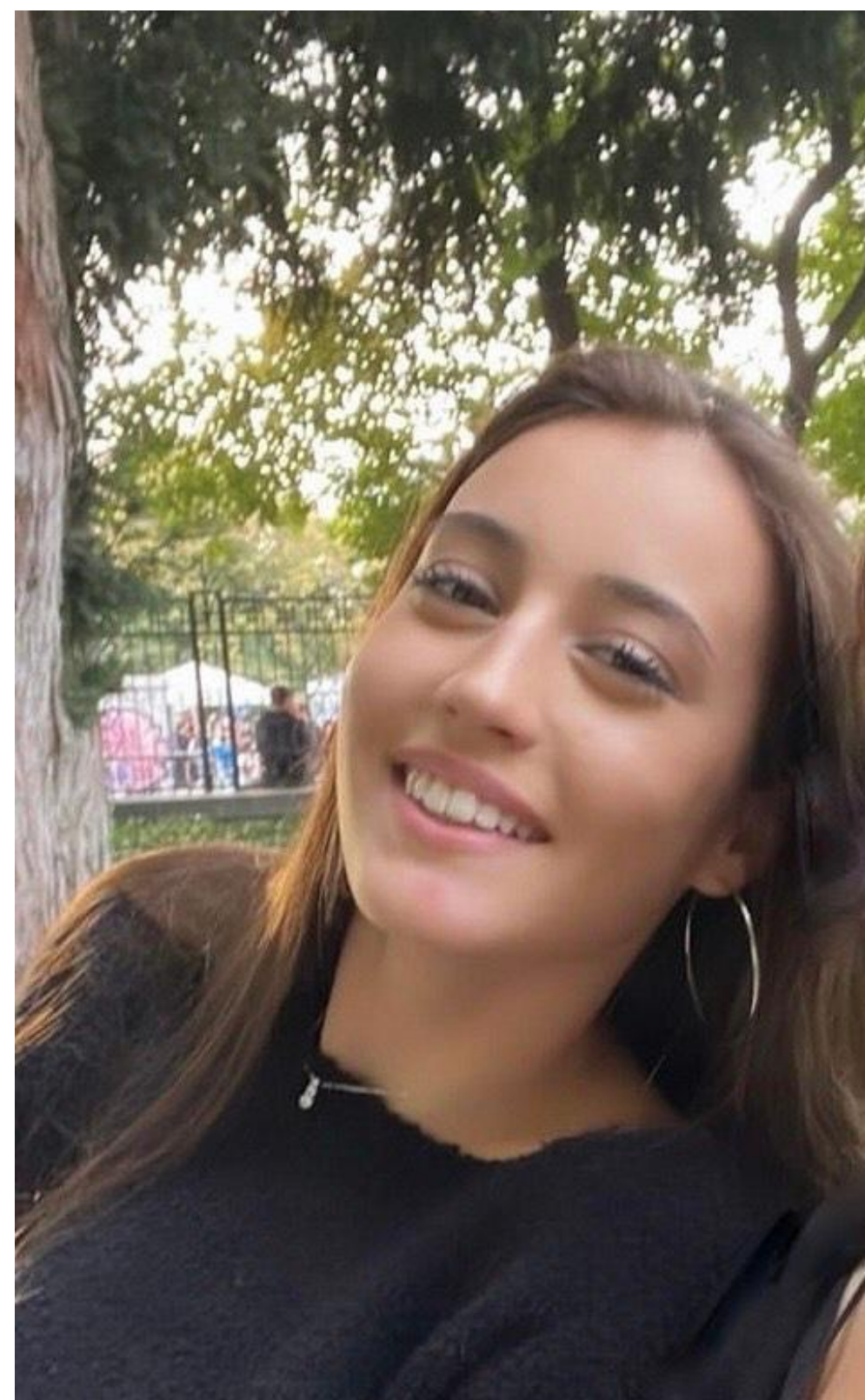
## WP6 Education and Training

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



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the European Union

## WP6 Team



Kami Markovska, Project Manager responsible for:

- Webinars
- Accreditation
- Clinical Exchange Programme
- Endorsements

Email: [kamelia.markovska@mu-varna.bg](mailto:kamelia.markovska@mu-varna.bg)

## WP6 Team



Sevim Zekerie, Project Assistant responsible for:

- Webinars
- Accreditation
- Clinical Exchange Programme
- Endorsements

As aid and at times replacement to the Project Manager and WP6 Chair

Email: [kamelia.markovska@mu-varna.bg](mailto:kamelia.markovska@mu-varna.bg)

## The 3 pillars of the Endo-ERN Academy

### Educational ERN Specific Webinars

#### Endo-ERN webinars

- Minimum 2 webinars per MTG per year
- “How to prepare a guideline”
- Direct invitations to authors of interesting papers
- Development of webinars from the ePAGs

#### Create workshop programme

- Roadmap of webinars in collaboration with ESE&ESPE RD Committees

### Joint webinar programme with ESPE & ESE

#### Extension of current endeavour in ESE Talks series:

- WP Evaluation
- WP4 Healthcare & CPMS
- WP7 CPGs & CDSTs.

### Endo-ERN relevant national/international symposia

ESE and ESPE: Symposia on rare endocrine disease at international conferences

National workshops/symposia run by ERN members and supported by ERN e.e. via dissemination, promotions, organization, facilitation, etc

## Webinar programme

### Endo-ERN webinars

- MTGs organise 2 to 3 webinars per year
- Use the [form](#) on the website to communicate information about the webinar

### ESE talks...webinars

- 5 Nov: [Thyroid hormone resistance](#)

### ENDO Connect webinars

- Oct 17: [Rare Thyroid conditions](#)

## 2024 webinars to date

MTG	Webinar
8	The differential diagnosis of discrepant thyroid function tests
1/7	Gonadal dysfunction in males with CAH
1/7	Pregnancy management in CAH
6	SOD (septo-optic dysplasia) Clinical Management and Patient Needs
3	The mysterious links among cilia centrosomes and insulin resistance: What have we learned from human genetics?
4	Multiple Endocrine Neoplasia Type 2: New insights from childhood to adult
2	Fibrous dysplasia with emphasis on hypophosphatemia
7	Turner syndrome and fertility
3	SWEET – the role of worldwide registry to investigate rare diabetes
7	The new genetics of Turner syndrome
5	Management of genetic obesity and food seeking behaviour e.d. in BBS/ALSTRÖM/PRADER WILLI syndromes / use of GLP-1 agonists in these conditions

## 2025 webinars

MTG	Webinar
7	Long-term outcomes in CAIS
5	Update SGA, GA treatment beyond childhood

- Please pre-plan your webinars for the first half of 2025 by **December 15** so we can apply for accreditation in a cost effective manner.

To ran a webinar on behalf of Endo-ERN is a **bonus** in the HCP Performance system

## Accreditation

- Accreditation received for 5 webinars in 2024
- Recording is accredited for 6 months after the event



## Symposium at ESPE 2024



Sunday, 17 November 1430-1600 in Hall 3

### *Future of Registries for Rare Disease: the example e-REC*

Erica van den Akker, Erasmus MC Children's Hospital, Rotterdam, the Netherlands



### *Genetic testing in DSD or rare forms of adrenal insufficiency - what is the relevance for therapeutic decisions?*

John Achermann, Great Ormond Street Hospital, London, UK



### *The patient perspective on new treatment options for hypogonadotropic hypogonadism*

Nils Smith, patient representative, UK

**Another bonus** – presentation of own HCP active research relevant to your expertise/MTG at conferences for rare diseases

## Endo-ERN Endorsement for educational events



- 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

# ENDORSED BY



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Endo-ERN

<https://endo-ern.eu/members/endorsement/>

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Webinar Planning

Termination Policy

## Endorsement of Educational Activities

Endo-ERN, via Work Package 6 (WP6), can **endorse educational activities** that are organised by institutions, national and international expert committees. Applicants are requested to complete the [endorsement form](#) and provide the requisite supporting documentation and [email](#) to the Endo-ERN Project Office.

### The criteria for endorsement is:

- activity concerns (a subgroup of) rare endocrine conditions
- activity provides both the paediatric and adult perspective of the condition(s) covered by the programme
- activity is organised by one or more than one Endo-ERN expert centre
- activity is accessible for healthcare providers and patient representatives from all EU member states
- condition(s) of the activity is/are covered within one of the eight Main Thematic Groups (MTGs)

## Endorsement Application Form



Please complete this form and return with requested documentation to the Endo-ERN project office at [endoern@amsterdamumc.nl](mailto:endoern@amsterdamumc.nl). Please make your endorsement request at least 6 weeks in advance. Endo-ERN will submit for endorsement activities that meet the requirements including but not limited to multiday conferences, workshops or webinars. **If you receive endorsement you are required to send an anonymized attendee report to Endo-ERN including total number of attendees and the country where they practice.**

**Requester name and email address:**

**Affiliated Endo-ERN Institution:**

Endo-ERN reference centre: patient organization, or partner organization)

**Event Name:**

**Event Date:**

**Event Location:**

**Does the programme cover:**  paediatric  adult  both

Endo-ERN members are encouraged to develop programmes that include sessions dedicated to adult and paediatric conditions or transition.

**Please indicate by ticking the box that your activity meets the criteria for an Endo-ERN endorsement:**

- activity concerns (a subgroup of) rare endocrine conditions*
- activity provides both the paediatric and adult perspective of the condition(s) covered by the program*
- activity is organised by one or more than one Endo-ERN reference centre(s)*
- activity is accessible for health care providers and patient representatives from all EU member states*
- condition(s) of the activity is/are covered within one of the eight Main Thematic Groups*

**Please indicate the relevant MTG by ticking the box/es below:**

- MTG1 Adrenal**  **MTG2 Disorders of Calcium & Phosphate Homeostasis**  **MTG3 Genetic Disorders of Glucose & Insulin Homeostasis**
- MTG4 Genetic Endocrine Tumour Syndromes**  **MTG5 Growth & Genetic Obesity Syndromes**  **MTG6 Hypothalamic and Pituitary Conditions**  **MTG7 Sex Development & Maturation**  **MTG8 Thyroid**

**Is this event accredited?**  YES  NO

**If yes, by what accreditation body?**

**If there is pharmaceutical sponsorship at this event please confirm the sponsor input:**

- sponsor had input into the programme
- sponsor had no input into the programme

**Please include the following information to complete your application by providing the appropriate links/document:**

- Website link to final agenda/programme or attach (please ensure the submitted programme is in English)
- Website link for registration or please describe below
- Logos and/or images for social media dissemination
- If there is pharmaceutical sponsorship for this event please indicate

**NB:** Endo-ERN requires a post-event the submission of total attendees broken down by country



Digital form coming soon!



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## Clinical Exchange Programme



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*Exchange Programme*

- 8 exchanges planned per year within the current grant
- **2025 call open from today until Nov 30th 2024** for Exchanges to be completed in **2025**
- Several members have volunteered to host and information is available on the [website](#).

Hosting is **expected** and participating is a **bonus** in the HCP Performance system

## 2024 Clinical Exchange Programme

Four approved exchanges for 2024 ongoing:

- Mater Dei Hospital Malta -> Beaumont & Children's Health Ireland
- HUS Helsinki University Hospital -> Ludwig Maximilian University Munich
- IRCCS Ospedale Policlinico San Martino -> Leiden University Medical Center
- M Sklodowska – Curie National Research Institute of Oncology, Gliwice Branch -> Instituto Auxologico Ospedale San Luca



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# WP7 Clinical Practice Guidelines & Clinical Decision Support Tools

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



Funded by  
the European Union

## Work package members

Olaf Dekkers **Adult chair**

Leiden University Medical Center (Leiden , Netherlands)



Anna Nordenström **Paediatric chair**

Karolinska University Hospital (Stockholm, Sweden)



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)

## Main goal WP7:



- To develop EndoERN clinical practice guidelines and decision-making tools for identified subject areas. Focus on knowledge gaps and guideline needs.
- Key considerations include:
  - Covering the transition from pediatric to adult endocrinology
  - 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.
- Review and update endorsed / appraised guidelines (MTG's will be asked to provide info)

## Pubertal induction and transition to adult sex hormone replacement in patients with congenital pituitary or gonadal reproductive hormone deficiency: an Endo-ERN clinical practice guideline

A Nordenström<sup>1</sup>, S F Ahmed<sup>2</sup>, E van den Akker<sup>3</sup>, J Blair<sup>4</sup>, M Bonomi<sup>5,6</sup>, C Brachet<sup>7</sup>, L H A Broersen<sup>8</sup>, H L Claahsen-van der Grinten<sup>9</sup>, A B Dessens<sup>10,11</sup>, A Gawlik<sup>12</sup>, C H Gravholt<sup>13,14</sup>, A Juul<sup>15,16</sup>, C Krausz<sup>17</sup>, T Raivio<sup>18</sup>, A Smyth<sup>19</sup>, P Touraine<sup>20,21</sup>, D Vitali<sup>22</sup> and O M Dekkers<sup>23,24</sup>



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*European Journal of Endocrinology*, 2024, **190**, G1–G14

<https://doi.org/10.1093/ejendo/lvae041>

Advance access publication 4 April 2024

Clinical Practice Guideline



## Familial hyperaldosteronism: an European Reference Network on Rare Endocrine Conditions clinical practice guideline

Paolo Mulatero,<sup>1,\*</sup> Ute I. Scholl,<sup>2</sup> Carlos E. Fardella,<sup>3</sup> Evangelia Charmandari,<sup>4,5</sup> Andrzej Januszewicz,<sup>6</sup> Martin Reincke,<sup>7</sup> Celso E. Gomez-Sanchez,<sup>8,9</sup> Michael Stowasser,<sup>10</sup> and Olaf M. Dekkers<sup>11</sup>

## Clinical Practice Guideline

A Nordenström and others

Pubertal induction: a clinical guideline

186:6

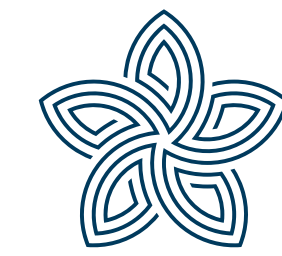
G9-G49

# Pubertal induction and transition to adult sex hormone replacement in patients with congenital pituitary or gonadal reproductive hormone deficiency: an Endo-ERN clinical practice guideline

A Nordenström<sup>1</sup>, S F Ahmed<sup>2</sup>, E van den Akker<sup>3</sup>, J Blair<sup>4</sup>, M Bonomi<sup>5,6</sup>, C Brachet<sup>7</sup>, L H A Broersen<sup>8</sup>, H L Claahsen-van der Grinten<sup>9</sup>, A B Dessens<sup>10,11</sup>, A Gawlik<sup>12</sup>, C H Gravholt<sup>13,14</sup>, A Juul<sup>15,16</sup>, C Krausz<sup>17</sup>, T Raivio<sup>18</sup>, A Smyth<sup>19</sup>, P Touraine<sup>20,21</sup>, D Vitali<sup>22</sup> and O M Dekkers<sup>23,24</sup>



European Reference Network

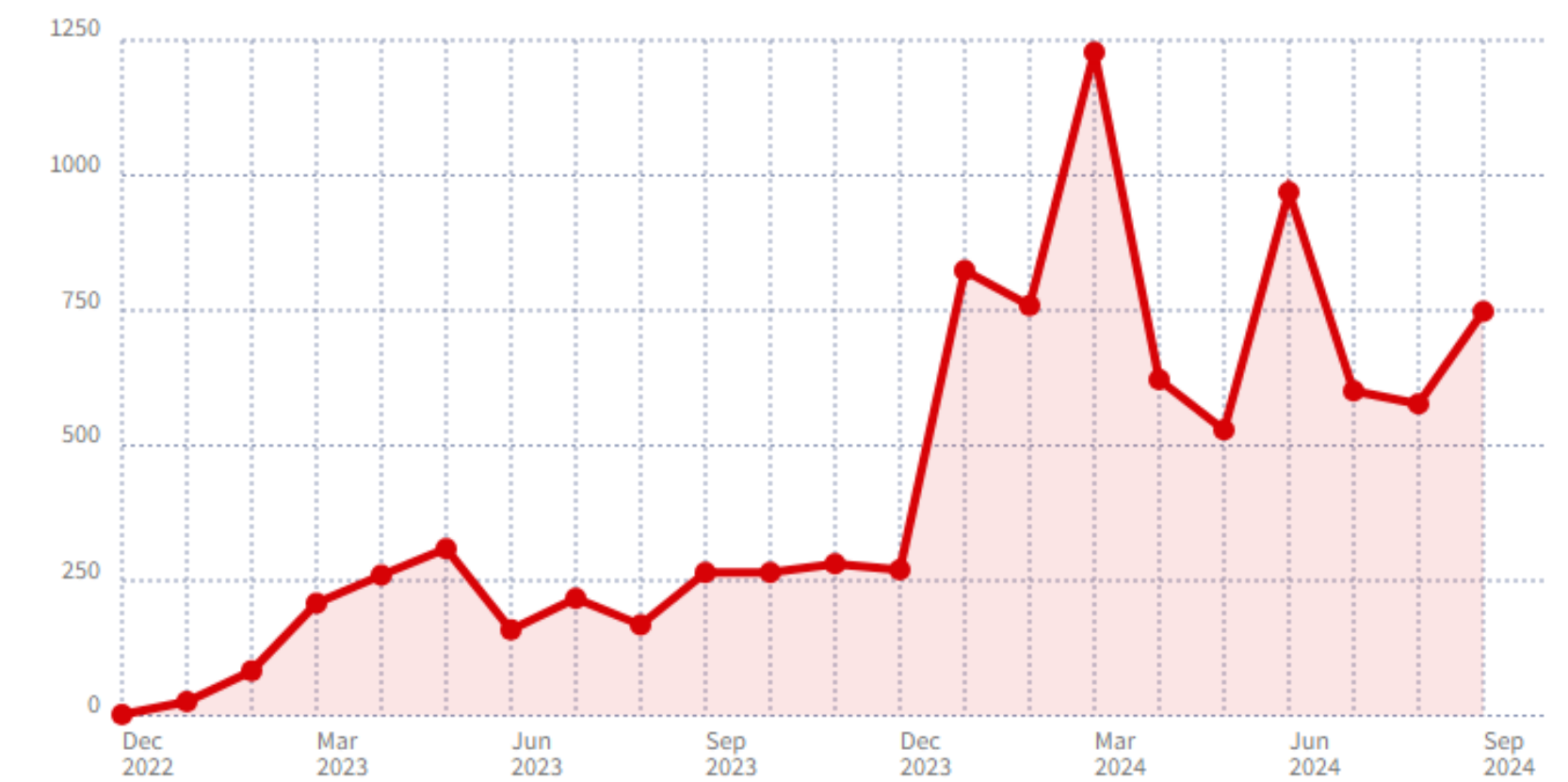


Endo-ERN

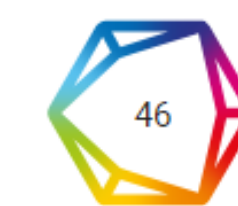
### Metrics

Total Views	6,559 Pageviews
9,370	2,811 PDF Downloads

Since 12/1/2022



### Citations



46 Total citations  
40 Recent citations  
35 Field Citation Ratio  
7.81 Relative Citation Ratio

Powered by Dimensions

### Altmetrics



13 Referenced in 1 policy sources  
Posted by 12 X users  
Referenced in 1 Wikipedia pages  
71 readers on Mendeley

26 Web of Science

# Guideline publications and plan



## AIM:

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

**2nd Guideline on Familial Hyperaldosteronism**, Published April 2024, presented ECE May 2024

**3rd Guideline on Transition in collaboration with MTG 6**, initiated

Patient involvement is a priority in the guideline development process.

Efforts will be made to:

- Create lay versions, to facilitate patient access and understanding.
- Translate guidelines into multiple languages and

Dissemination strategies will include:

- CME credits, downloadable slide decks etc

# Endorsement



- What are your opinions regarding endorsement?
  - What to endorse?
    - Guidelines? Consensus statement? Expert opinion document?
  - When to endorse?
    - Endorse when ENDO-ERN was (formally) involved?**
    - Routinely endorse all guidelines from neighbour societies?
  - Who should endorse?
    - MTG's?**
    - WP7
    - The ENDO-ERN board?

Process of endorsement needs to be formalized.

## For discussion

1. How does the transition in care (Prof Isidori) general guideline relate to the current transition in pituitary care guideline?
2. What's the next topic? (Good to initiate the next guideline)



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## WP8 & WP9 Activities for Ukraine

Alberto Pereira, Adult Chair, Endo-ERN



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# Other and discussion

## Meetings

- 9th Endo-ERN General Assembly – Friday 9 and Saturday 10 May 2025, Copenhagen, Denmark
- Next (online) SCAB meeting: to be scheduled in February 2025
- MTG specific meetings for HCPs

MTG1: 27 November, 15-16 hrs

MTG2: **TBD**

MTG3: 23 October, 16-17 hrs

MTG4: **TBD**

MTG5: 7 October, 15-16 hrs

MTG6: **TBD**

MTG7: 30 October, 17-18 hrs

MTG8: 12 November, 16-17 hrs

## Meetings



ERN Research Conference, Udine, Italy

Dear ERN Research community,

**Attention deadline approaching! Call for Abstracts related to 'ERN Research' ([call text](#))**

And don't forget to register to:

**European Rare Disease Research Coordination and Support Action- ERICA  
ERN Research Conference**

**Wednesday 11 December (14:00 CEST Opening) - Friday 13 December 2024**

Hosted by Azienda Sanitaria Universitaria Friuli Centrale Udine / MetabERN  
Venue: Hotel Là di Moret, Udine, Italy

All the ERICA stakeholders, ERN representatives, Expert Group members, Advisors and partners will gather to this face-to-face symposium to discuss the progress and future of the ERN related Research activities. We call all the stakeholders to think What is ERN research or what should be typical ERN research?

Great opportunity to meet the active ERNs research community and to shape the future of the RD Research!