

European Registries For Rare Endocrine Conditions

EuRRECa

eurreca.net













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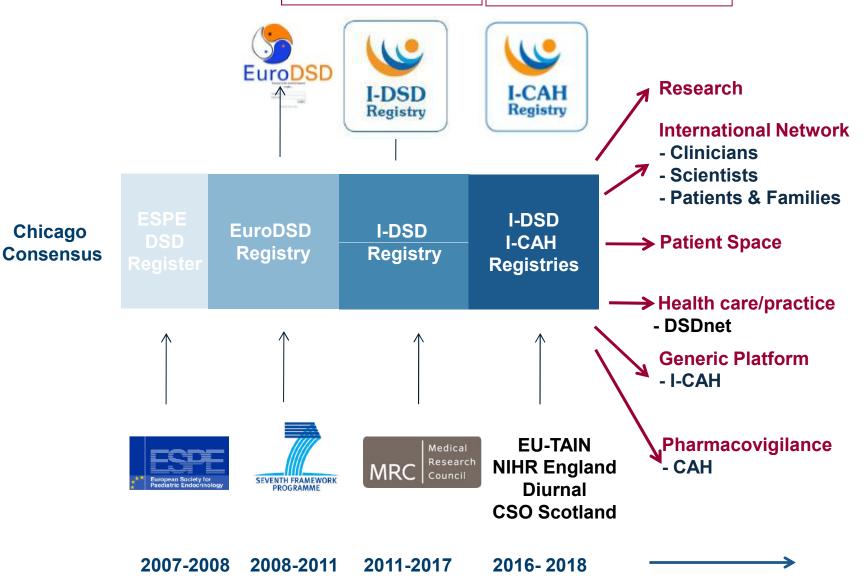


Registries For Conditions Affecting Sex Development

Project Group

Anthony Stell, Richard Sinnott, Faisal Ahmed

Jipu Jiang, Jillian Bryce, Martina Rodie, Richard Sinnott, Faisal Ahmed Jillian Bryce, Sam Akanimo, Salma Ali, Tom Muir, Hannah Van Hove, Office For Rare Conditions, Faisal Ahmed





International Detailed Disease Registries

- Uniform platform for collecting data





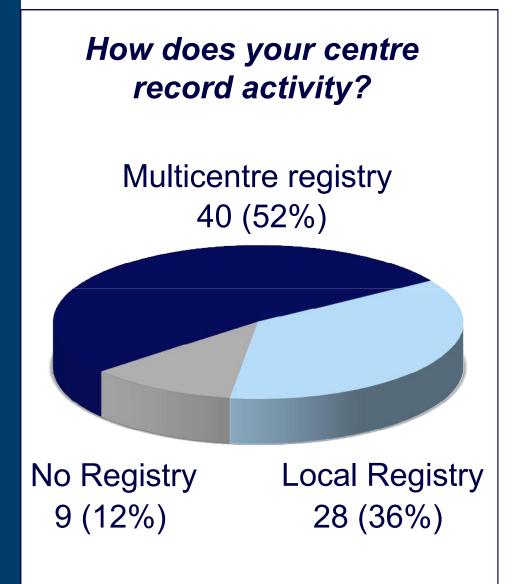


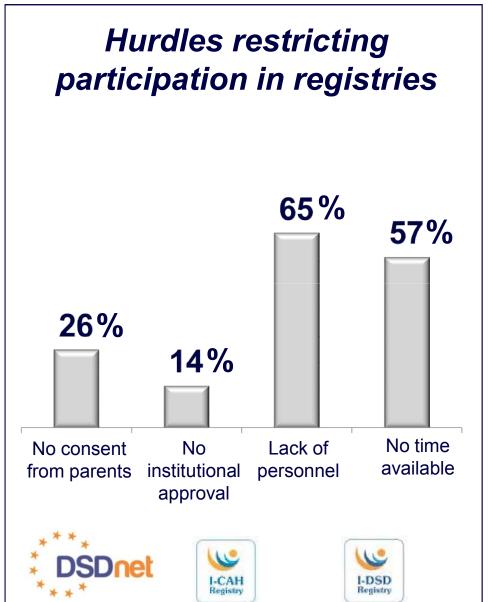
>3000 cases

Newborn to 90yrs Median Age – 16 yrs Approved by ethics to store routinely collected clinical data following informed consent



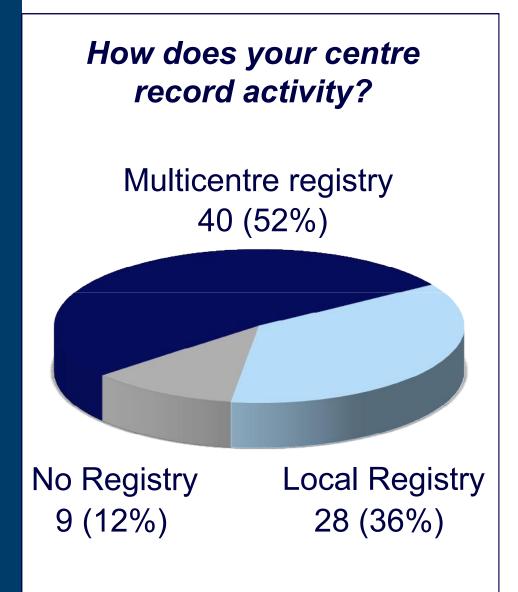
Participation In DSD Registries

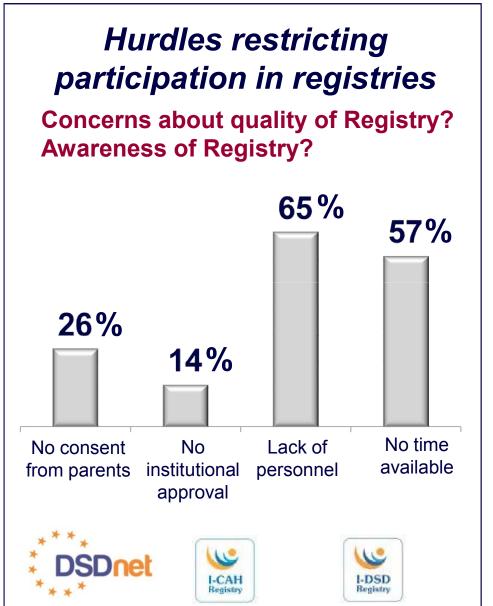






Participation In DSD Registries







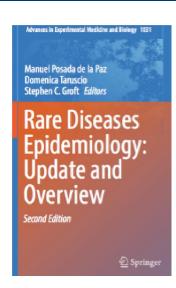
There Is A Science To Registries!

Chapter 8 Data Quality in Rare Diseases Registries

Yllka Kodra, Manuel Posada de la Paz, Alessio Coi, Michele Santoro, Fabrizio Bianchi, Faisal Ahmed, Yaffa R. Rubinstein, Jérôme Weinbach, and Domenica Taruscio

In Europe – > 600 Rare Disease Registries

local, national and international



- Concerns about quality, sustainability, codification and interoperability

Kourime et al. Orphanet Journal of Rare Diseases (2017) 12:56 DOI 10.1186/s13023-017-0603-7 Orphanet Journal of Rare Diseases

RESEARCH

Open Access



An assessment of the quality of the I-DSD and the I-CAH registries - international registries for rare conditions affecting sex development



Information Quality

Some Components Of Quality

Networking Function

Research Quality Evidence Quality Level of Activity - Registry Design - Enrollment of centres - Internal Validity - Operational Aspects - Rate of addition of cases - External Validity - Data Quality Quality - Completed Studies - Role in the network - Scope of data sharing - Active Studies - Patient involvement - Origin of centres - Types of studies

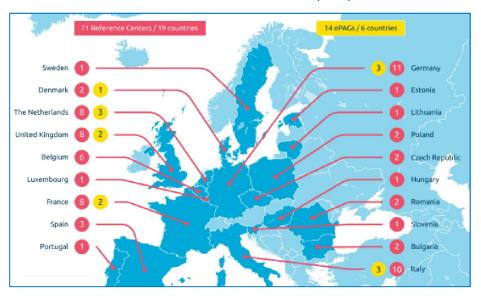
Many of these components are dependent on Awareness of Registry

International Acceptability

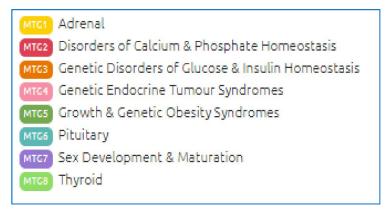


- European Reference Networks (ERN) clinical networks that aim to manage rare conditions requiring specialist input and a concentration of knowledge and resources
- ERN for rare endocrine conditions (Endo-ERN) largest ERN and includes 36 groups of conditions

71 Reference Centres (RC)



8 Thematic Groups



- The range of cross-border registries that exist for Endo-ERN conditions is unclear
- The creation of Endo-ERN provided an opportunity to study the existence of registries in Europe



Assessing The Awareness Of Registries

Database search for international registries using Orphanet and RD-Connect using Orphacodes for all rare endocrine conditions within Endo-ERN

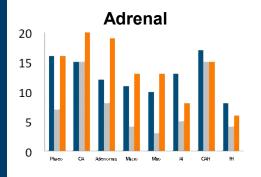
> •An 'international registry' was defined as that used by >1 country with the coordinating centre based in Europe

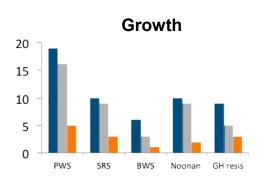
A Survey of 71 RC from 19 countries within Endo-ERN (Oct-Nov 2016):

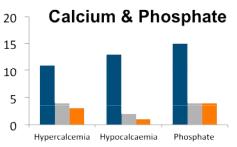
- Awareness and participation in registries at an international, national and local level for Endo-ERN conditions within main thematic groups (MTG)
- Views on future priorities for new registries for conditions within each MTG

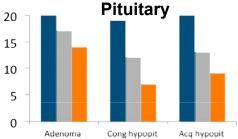


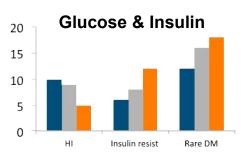
Awareness of Endo-ERN Members In Registries

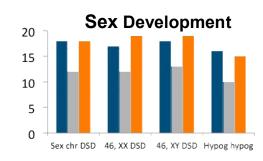


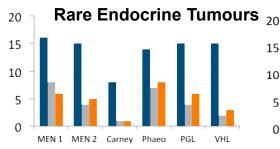


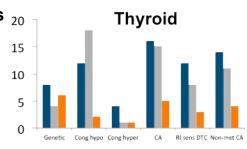










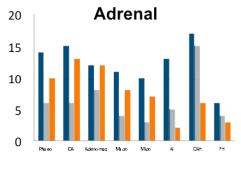


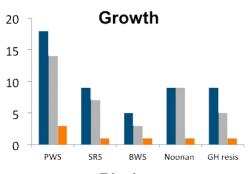


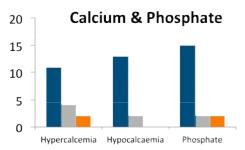
- Greatest awareness of international registries- MTG1 (Adrenal), MTG3 (Glucose & Insulin), MT7 (Sex Dev)
- Lowest awareness of international registries- MTG2 (Ca & Phos), MTG4 (Endo Tumours), MTG5 (Growth), MTG6 (Pituitary), MTG8 Thyroid
- High levels of awareness of national and local registries

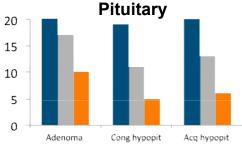


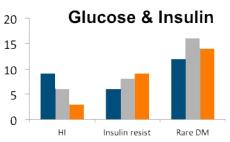
Participation of Endo-ERN Members In Registries

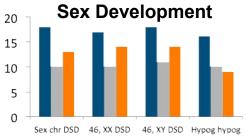


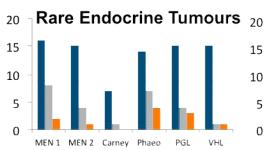


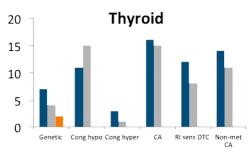














- Greatest participation in international registries for conditions within MTG1 (Adrenal), MTG3 (Glucose & Insulin), MT7 (Sex Dev)
- Despite awareness of registries, participation rates lower
 - e.g. 70% 'aware' of an international registry for DSD, yet, participation rate was 52%
- No participation in international registries for some conditions e.g. MTG8 (Thyroid)

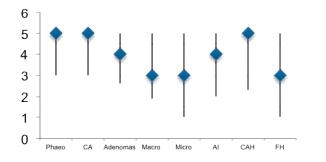


Views On Future Priorities

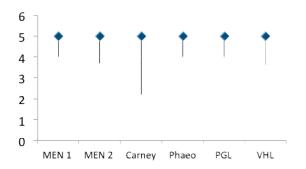


Priority (median, 10th 90th)

◆ MEDIAN

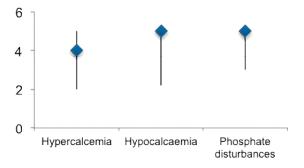


Rare Endocrine Tumours

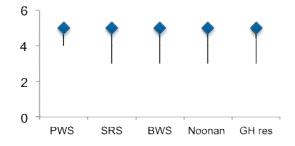


- A score of 5 was attributed to all conditions within:
- MTG4 (Rare Endocrine Tumours)
- MTG5 (Growth & Obesity)
- MTG7 (Sex Development)

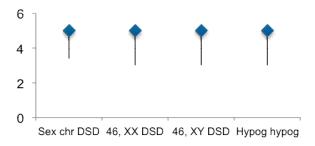
Calcium & Phosphate



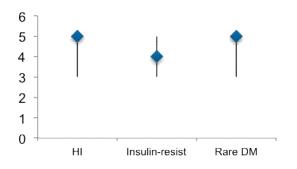
Growth & Obesity



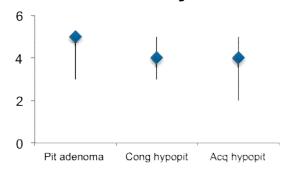
Sex Development



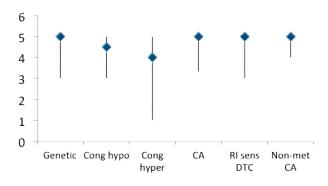
Glucose



Pituitary



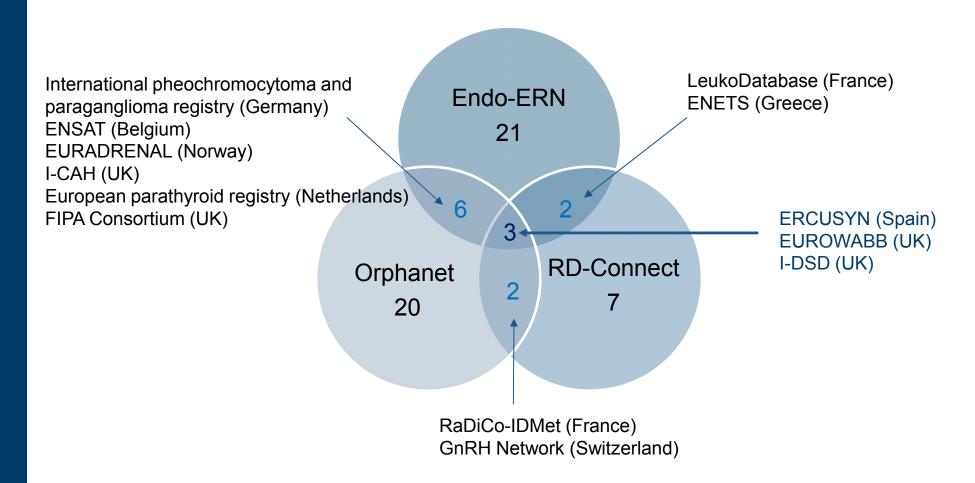
Thyroid





International Registries For Rare Endocrine Conditions

- Registries exist for 76% (32/42) of Endo-ERN conditions
- Several registries including all commercial registries not present in databases of registries such as Orphanet & RD-Connect





- Several international registries exist for a range of conditions
- Still gaps in the coverage of conditions that needs addressing
- Bigger gaps in the levels of awareness and use of existing registries
- 'Registry libraries' such as Orphanet and RD-Connect do not have a record of several international registries led from Europe

The current landscape of European registries for rare endocrine conditions

in European Journal of Endocrinology

Authors: S R Ali ^{1, 2}, J Bryce ², M Cools ^{3, 4}, M Korbonits ⁵, J G Beun ⁶, D Taruscio ⁷, T Danne ⁸, M Dattani ⁹, O M Dekkers ¹⁰, A Linglart ¹¹, I Netchine ¹², A Nordenstrom ¹³, A Patocs ¹⁴, L Persani ^{15, 16}, N Reisch ¹⁷, A Smyth ², Z Sumnik ¹⁸, W E Visser ¹⁹, O Hiort ²⁰, A M Pereira ²¹, S F Ahmed ^{1, 2} and on behalf of Endo-ERN



December 2016 Endo-ERN Registries Workshop











The Case For EuRRECa

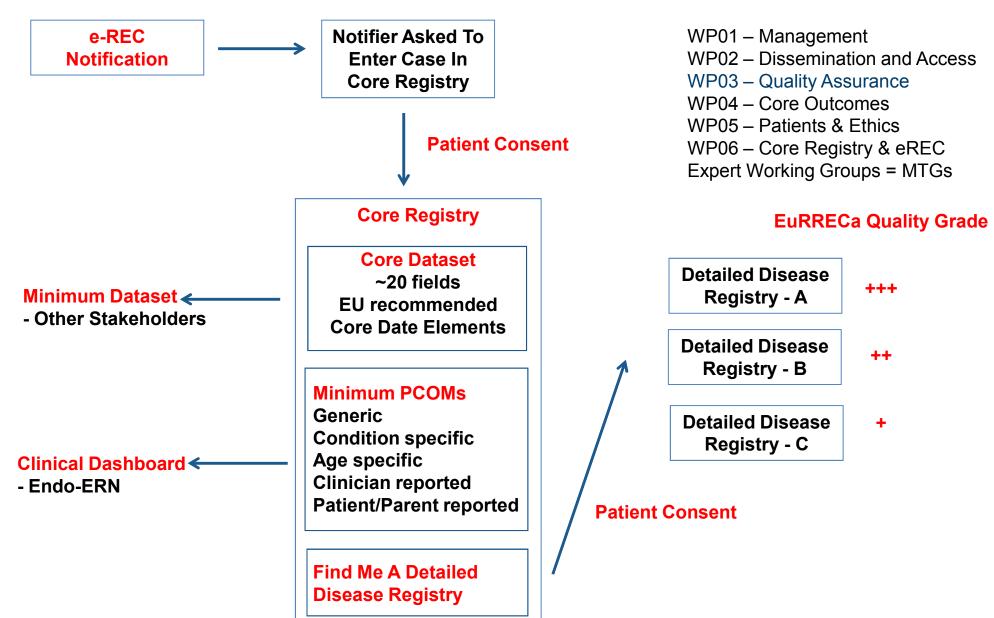
- A need to develop new registries for rare endocrine conditions
- A more immediate need to improve the awareness and participation in existing registries
- A core registry which directs users to high quality detailed registries is required



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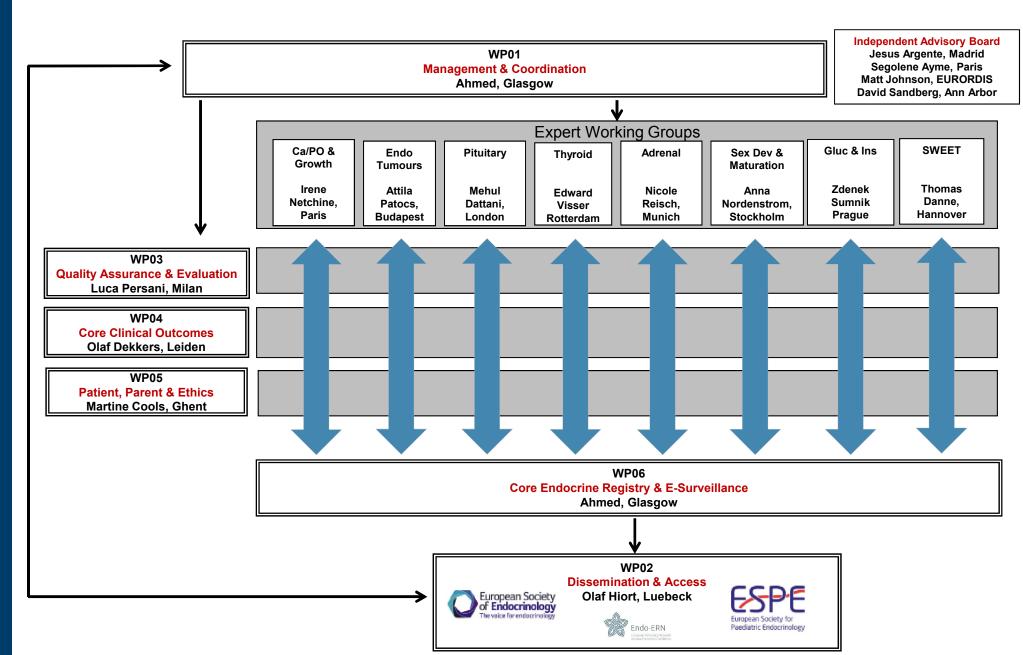


The Concept Of EuRRECa





The Organization Of EuRRECa





The Review Of The Application By EU

- Added value to the field of public health is significant primarily due to the effectiveness and efficiency of the proposed method and the integration across several key stakeholders. The considerations for linguistic diversity is ambitious and shows a patient centeredness, which should lead to better access for more patients.
- Project work is well integrated in the 71 member ERN, which together with the involvement of European professional associations provides for an excellent geographic coverage. The social, cultural and political context are generally well addressed, including the potential impact of Brexit on the project
- Consortium provides a thorough account of the current challenges, open issues and state- of the-art in a comprehensive and wide-span fashion. A detailed analysis with diagnosis is provided of the present situation of registries. The elements for improvement are identified and are at the basis of the proposed content of the project.
- Described methodology addresses appropriately interoperability and secures a standards-based development of the core Registry. There is good complementarity with the ENDO ERN activities while the approach is promising as to the integration/linking of existing relevant registries in a transparent and accountable way.
- Work plan is very well designed and efficient with proper identification of tasks. The management structure and internal communication are appropriate. The different bodies, their role and composition is well described. The embedding of the project in the ERN is also an advantage for internal communication.
- Partnership is of high quality and the strong link with the ERN and two professional societies is embedded in the partnership.







Launched in Feb 2018 - Activity To Date

EuRRECa Newsletter July 2018

EuRRECa (European Registries for Rare Endocrine Conditions) is supported by the EU Health Programme, the European Society for Paedistric Endocrinology (ESPE) and the European Society of Endocrinology (ESPE). The project which is closely linked to the European Reference Network for Rare Endocrine Conditions (Endo-ERN) was launched in February 2018 and aims to support the needs of the wider endocrine community.

Aim Of The Newsletter

The aim of the newsletter is to provide an update on recent activities. The newsletter will be issued twice a year at the end of January and July. You can subscribe to the newsletter at <u>eurreca.net/newsletter</u>.

EuRRECa in Brief

Specific aims of the EuRRECa project include:-

- e-REC: g-reporting programme of <u>Rare Endocrine</u> Conditions
- Develop a new Core Endocrine Registry that collects a core dataset for all rare endocrine conditions in Endo-ERN
 Incorporate core clinical outcomes in the Core Endocrine
- Registry
 Signpost participants to detailed, disease-specific
- registries that have been endorsed by EuRRECa
- Inform the development of new high-quality detailed disease registries

The EuRRECa Website

Detailed information is available at the project's new website <u>surrecanet</u>. You will find information on milestones, deliverables, the Work Packages and the Expert Working Groups that form the EuRRECa Project Group, the Independent Advisory Board, past and future events and also resources that are relevant for developing registries. So please visit the website and do let us know if you have any ideas of how it can be improved.

EuRRECa At ESE & ESPE 2018

The EuRRECa team was present at ESE 2018 in Barcelona at a joint stand with Endo-ERN and it will be present at ESPE 2018 in Athens. EuRRECa will also be prominent in the main programme (FC14 and ERN1.5).

Registries Kick off Meeting In Luxembourg



e-REC

e-REC (e-Reporting Of Rare Endocrine Conditions) is an electronic reporting system which captures activity and allows for a better understanding of the occurrence of rare endocrine conditions. This system will allow Endo-ERN to objectively map the conditions, the expert centres and other related activity. The reporting platform has been undergoing a limited pilot which will be followed by a more extensive pilot from August 2018. Although e-REC is currently aimed at serving the needs of Endo-ERN, it is open to all endocrine health care professionals. To find out more or to participate, please visit gurreca.net/e-rec.

Core Data Elements For The Core Endocrine Registry

WP3 on Quality Assurance & Evaluation and WP6 on Registry Development met in July at a meeting hosted by Luca Persani to discuss the core data elements (CDE). A draft copy is now available on the EuRRECa website.

Core Clinical Outcomes

Olaf Dekkers in WP4 has received valuable input from the EURRECa Expert Working Groups. A preliminary list of condition specific clinical outcomes can now be explored for future inclusion into the Core Endocrine Registry.

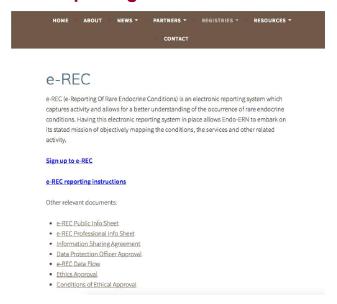
News From The Patient, Parents & Ethics WP

WPS on Patient Parent & Ethics led by Martine Cools met in Ghent to develop a set of recommendations that can guide the development of registries for rare endocrine conditions. The draft recommendations will soon be available for open consultation.

Recommendations On Rare Disease Registries

A group of experts, including members of EuRRECa, have recently published recommendations in the International Journal of Tonrisonmental Research & Public Health that can be used as a framework to guide the organizational and data quality of rare disease registries. These recommendations will guide the development of the toolkit that EuRRECa will use to support the quality of

- E-Reporting of Rare Endocrine Conditions



- Core Data Elements for the Core Endocrine Registry
- Core Condition-Specific Clinical Outcomes
- Draft Recommendations by Patients, Parents & Ethics







Parries

Recommendations for Improving the Quality of Rare Disease Registries

Yllka Kodra ^{1,*}, Jérôme Weinbach ², Manuel Posada-de-la-Paz ³ ³, Alessio Coi ^{4,5} ⁶,

S. Lydie Lemonnier ⁵, David van Enckevort ⁷ ³, Marco Roos ⁸, Annika Jacobsen ⁹ ³,

Ronald Cornet ⁹ ⁵, S. Faisal Ahmed ¹⁰, Virginie Bros-Facer ¹¹, Veronica Popa ¹²,

Marieke Van Meel ¹³, Daniel Renaull ¹⁴, Rainald von Gizycki ¹⁵, Michele Santoro ^{4,5},

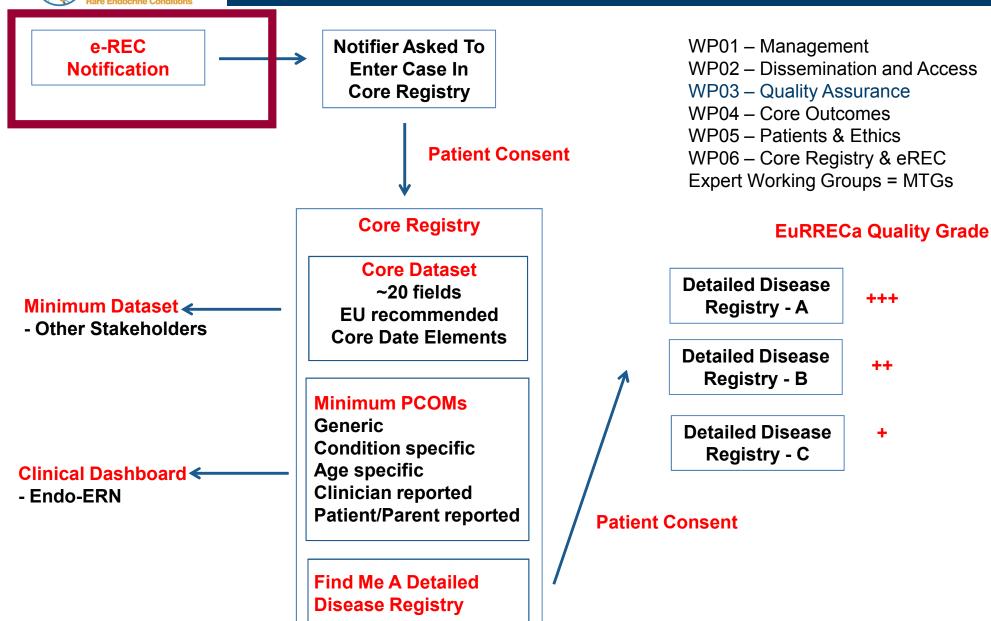
Paul Landais ^{2,16}, Paola Torreri ¹, Claudio Carta ¹, Deborah Mascalzoni ¹⁷, Sabina Gainotti ¹⁸ ⁵,

Estrella Lopez ³ ⁶, Anna Ambrosini ¹⁵, Heimo Müller ²⁰, Robert Reis ²⁰, Fabrizio Bianchi ^{4,5},

Yaffa R. Rubinstein ²¹, Hanns Lochmüller ^{22,28} and Domenica Taruscio ¹ ⁵



The Concept Of EuRRECa





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CONTACT
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e-REC

e-REC (e-Reporting Of Rare Endocrine Conditions) is an electronic reporting system which captures activity and allows for a better understanding of the occurrence of rare endocrine conditions. Having this electronic reporting system in place allows Endo-ERN to embark on its stated mission of objectively mapping the conditions, the services and other related activity.

Sign up to e-REC

e-REC reporting instructions

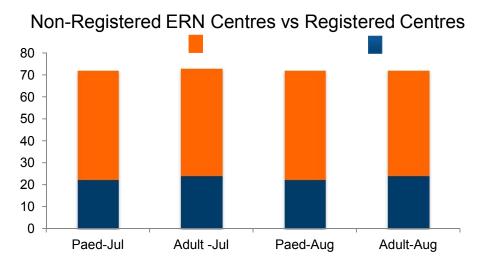
Other relevant documents:

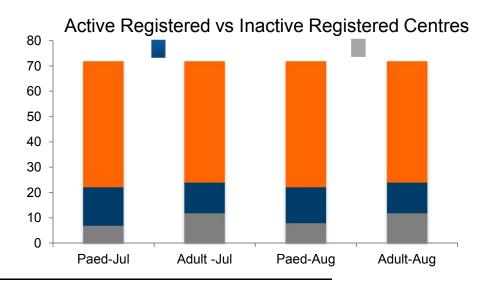
- · e-REC Public Info Sheet
- · e-REC Professional Info Sheet
- · Information Sharing Agreement
- Data Protection Officer Approval
- e-REC Data Flow
- · Ethics Approval
- · Conditions of Ethical Approval

ork needs to be ed into other nly used languages

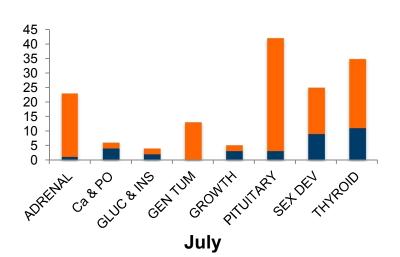


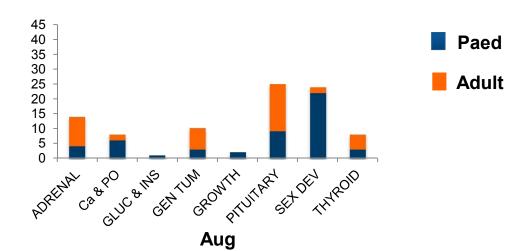
e-REC Update Of Pilot





	Paediatric	Adult
Number of people who are reporting in total	34	51
Number of people reporting per centre	1 (1, 7)	1 (1, 14)







Further Information & To Keep In Touch

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Home

EURRECa (European Registries for Rare Endocrine Conditions) which is funded by the European Union's Health Programme is almed at maximising the opportunity for all patients, health care professionals and researchers to participate and use high-quality, patient-centred registries for rare endocrine conditions covered within the European Reference Network on Bare Endocrine Conditions (Endo-ERN).

The project, launched in February 2018, will achieve its aim by running an e-reporting programme (e-REC), developing a new core endocrine registry that collects a core dataset that also includes objective markers of clinical outcome and, is astly, by signposting participants to high-quality, detailed, disease-specific and patient-centred registries that have been evaluated by EURRECs.

The project builds on the structure that has been created by Endo-ERN but will also be open to all health care professionals beyond this ERN. EURRECa receives guidance from Expert Advisory Groups that align with the <u>Main Thematic Groups</u> of Endo-ERN and their guidance will flow through work packages that will review the needs of patients, and parents, comply with the highest ethical standards, evaluate the quality and interoperability of datasets and combine them with patient-centred clinical outcomes. Clear policies that are acceptable to patients, researchers and industry for accessing data for research coupled with widespread dissemination and knowledge exchange through closely affiliated professional endocrine societies, patient support groups and across all the ERNs will ensure that EURRECa is sustained beyond the current lifetime of the project.













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- Exciting times for European endocrinology!
- Endo-ERN great example of how clinical centres across
 Europe can work together at improving patient care
- Effective but simple registries are a cornerstone of networks
- EuRRECa builds on Endo-ERN and aims to extend its reach further further across all centres within and beyond Endo-ERN
- EuRRECa aims to provide the evidence that networks for rare (endocrine) conditions can provide the infrastructure that can be used to improve clinical care and research