European Reference Network

Endo-ERN

ePAG & Coordinators meeting

Date: 09.09.2025

Time: 10:00 – 11:00 hrs

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Meeting agenda:

1. Opening and welcome (Charlotte)
2. New ePAGs Laura Maag and Marc Budenbender
3. Evaluation activities (Emily)
4. Patient journeys (Emily)
5. Collaborative project on drug shortages (Johan/Petra)
6. Communication Update (Aimee)
7. ePAG chairs: EURORDIS joint activities, other (Johan / Petra)
8. Other

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Opening and welcome

Charlotte van Beuzekom

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Introduction – new ePAG

Marc BÜDENBENDER
Let's Cure ACC, Member of the Board

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My Story …

Diagnoses

2017/07
- 20cm ACC tumor, local invasion/ infiltration, Ki67 20-30%, non-functioning

2019/04
- 5-6cm single liver metastasis, Ki67 60%

2019/07
- numerous liver metastases (up to 5cm)
- “palliative” situation

2024/07
- no evidence of disease

Treatments

- resection (R0)
- start Mitotane treatment
- 4 cycles EDP(M)

- resection (R0)
- 1st genetic analysis (mutated TP53)
- ongoing Mitotane treatment

- 6 cycles EDP(M)
- resection (R0) of remaining necrotic tumor in 2020/01
- ongoing Mitotane treatment

- stop of Mitotane treatment
- 2nd genetic analysis (no genetic predisposition)

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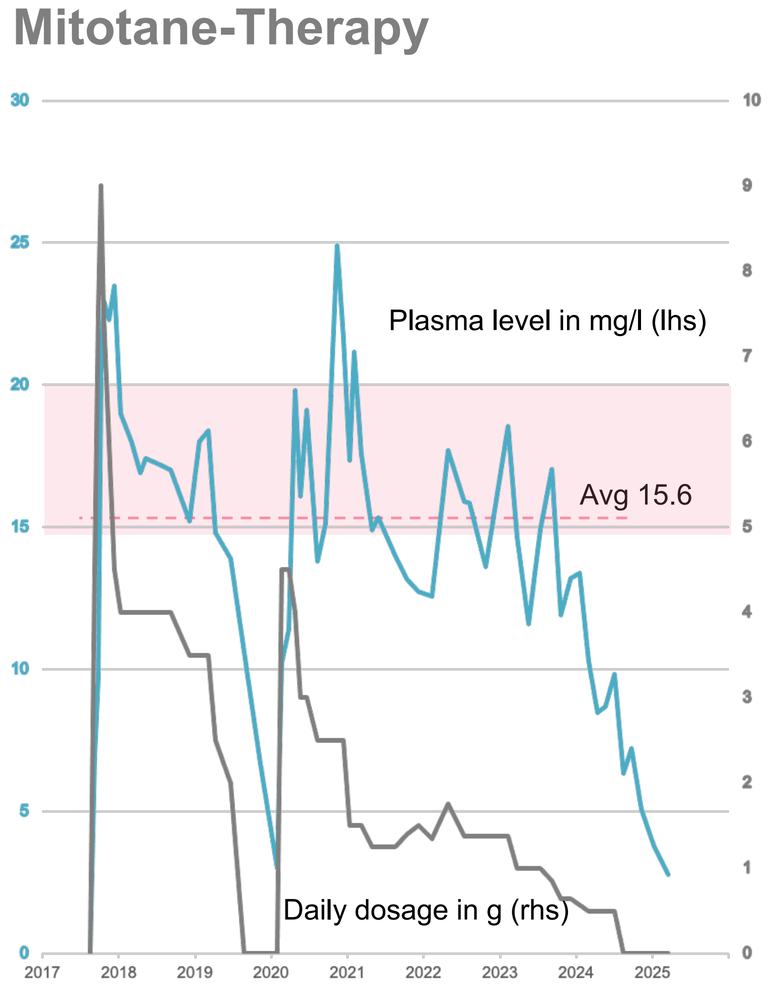

Overall, 9 Addison-Crises

By Patients for Patients

“Let’s Cure ACC aims to foster a strong, well-informed, and interconnected ACC community, ultimately offering patients the best support and striving for optimal outcomes.”

- Website with in-depth information in 11+ languages
- Around 200 one-on-one patient discussions & German Group in ‘24
- Database with 150+ experts in ca. 50 countries, incl. pediatric focus
- 13 Ambassadors worldwide speaking 10 languages

- Strong connections to the global ACC medical community
- Guided by our Medical Advisory Board:

- Guillaume Assié
- Kaitlin J. Basham
- Alfredo Berruti
- Tobias Else
- Martin Fassnacht
- Gary D. Hammer
- Electron Kebebew
- Katja Kiseljak Vassiliades
- Raul C. Ribeiro
- Cristina L. Ronchi
- Massimo Terzolo

Q&A

marc@letscureacc.com

www.letscureacc.com

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or #letscureacc

Evaluation activities

Emily White

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ePAG Activities Evaluation

1st Sept.

- Overview of activities sent to all ePAGs

19th Sept.

- Extension of deadline for comments/ confirmations

Current:

- Feedback received from several

20-30th Sept.

- Review of key areas of interest and work for Endo-ERN ePAGs with WPs
- Start refining list for January 2026 continuous monitoring

Thank you to those who have already responded!

Patient journeys

Emily White

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Patient Journeys - Update

June-September 2025: Endo-ERN adapted protocol for creating patient journeys piloted for first journey **Acromegaly**

Initial protocol from EURORDIS methodology requires specification to rare endocrine and our ERN needs.

Next slides go through the initial adapted protocol and lessons learned from our pilot

Lesson 1: Defining scope of Patient Journey needs additional engagement with MTG chairs ePAG & clinical – this PJ was an exception due to existing data gathered by our network

Why first patient journey

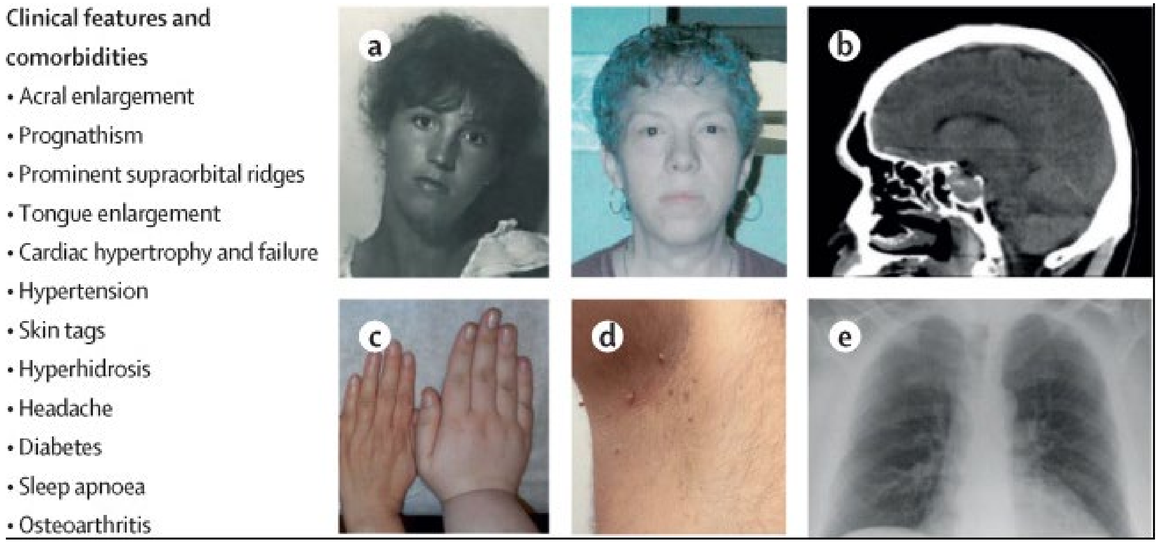
Relevant:

- Rare acquired endocrine disease
- Average time to diagnosis: 4 -10+ years

79 patient cases identified in Endo-ERN centres in 2023

63 expert centres in 23 MS in MTG6

Guidelines and experts exist within the Endo-ERN covering 24

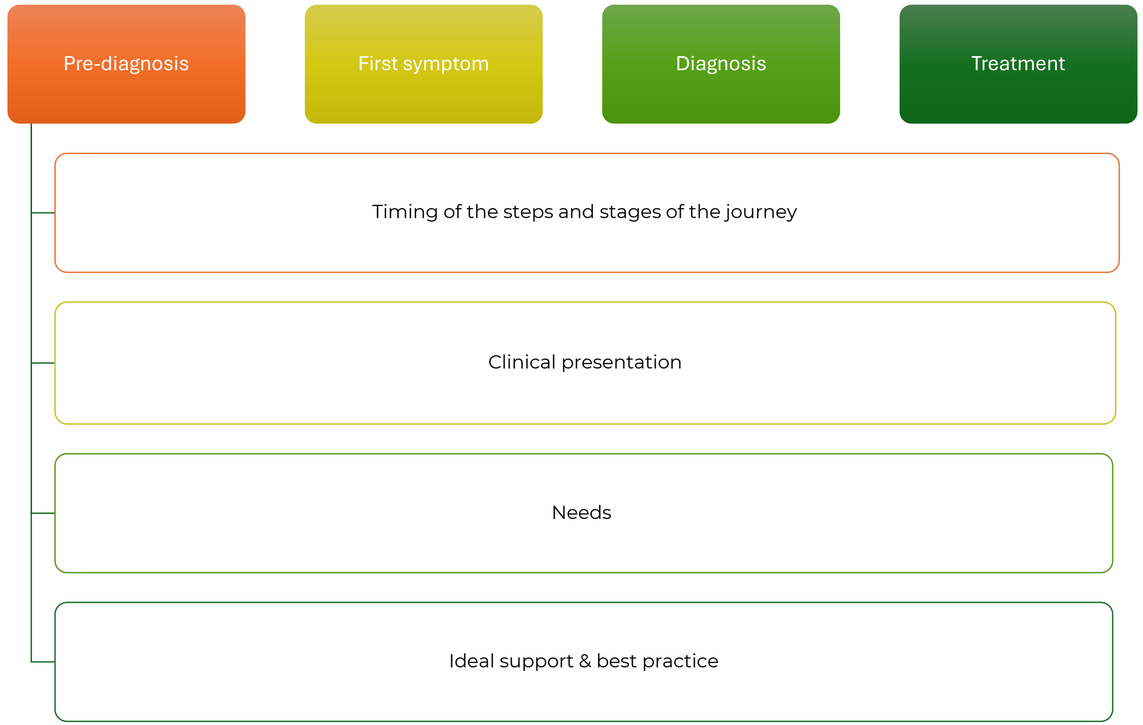


Chanson & Salenave, 2008
Fleseriu et al., 2022

Lesson 2: Minimum of 6 EU countries may not be possible for baseline creation – to achieve this for Final PJ, the timeline needs translation steps included for 6 languages

2. Define Stage of Journey

Min.= 20 patients , 6 EU countries (stratified sampling strategy)



Mixed methods analysis

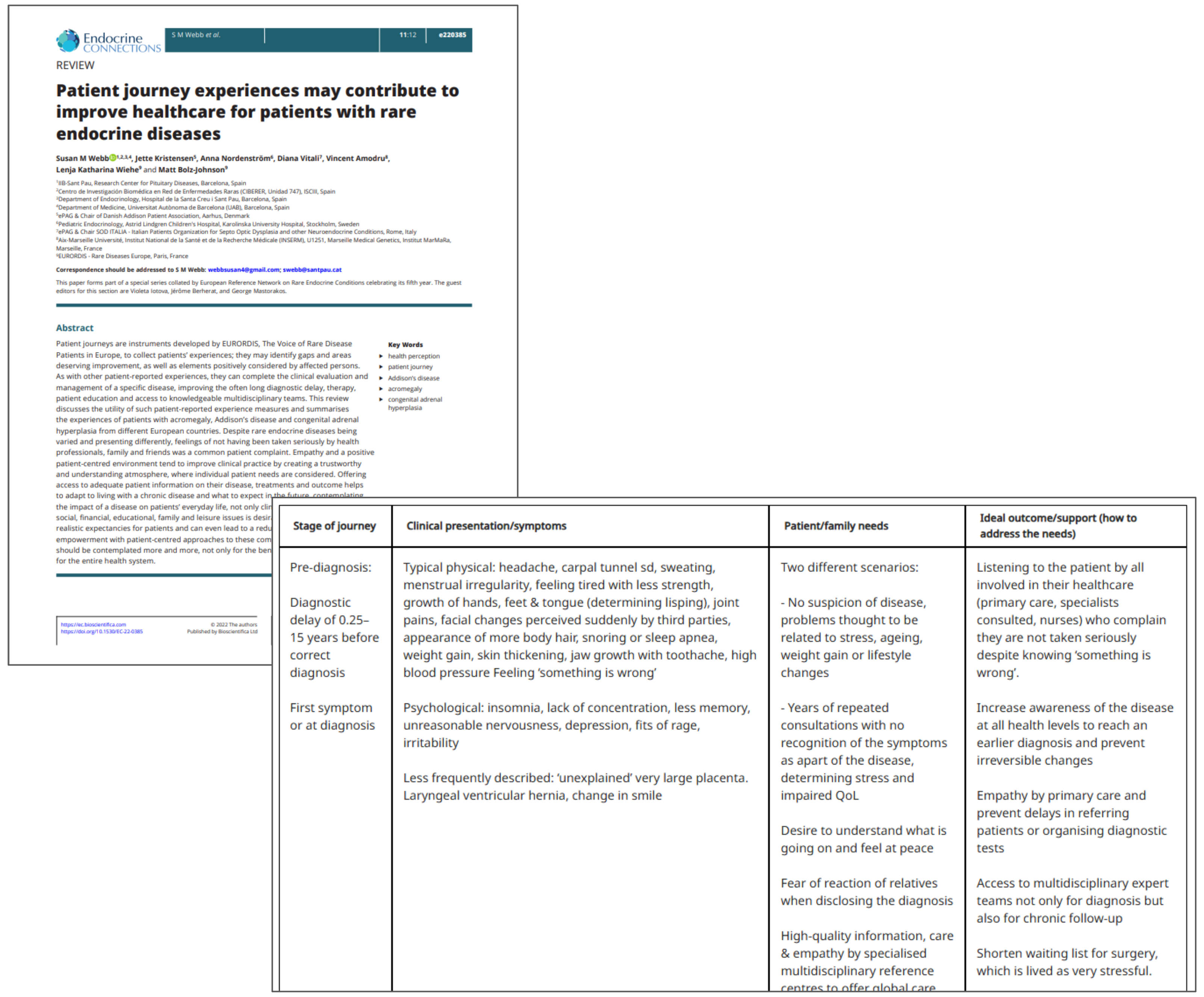
Inclusion criteria:

- 18 + years
- Residing in Europe
- Able to provide informed consent
- Able to communicate answers to questions themselves or through a trusted third party such as personal carer or legal guardian

Exclusion Criteria:

- Patients unable to communicate in one of the study languages.
- U18 yrs / current gigantism diagnosis

3. Engage with Community



Endocrine CONNECTIONS | S M Webb et al. | 11:12 | e220385

REVIEW

Patient journey experiences may contribute to improve healthcare for patients with rare endocrine diseases

Susan M Webb[1,2,3,4], Jette Kristensen[5], Anna Nordenström[6], Diana Vitali[7], Vincent Amodru[8], Lenja Katharina Wiehe[9] and Matt Bolz-Johnson[9]

[1]IIB-Sant Pau, Research Center for Pituitary Diseases, Barcelona, Spain
[2]Centro de Investigación Biomédica en Red de Enfermedades Raras (CIBERER, Unidad 747), ISCIII, Spain
[3]Department of Endocrinology, Hospital de la Santa Creu i Sant Pau, Barcelona, Spain
[4]Department of Medicine, Universitat Autònoma de Barcelona (UAB), Barcelona, Spain
[5]ePAG & Chair of Danish Addison Patient Association, Aarhus, Denmark
[6]Pediatric Endocrinology, Astrid Lindgren Children's Hospital, Karolinska University Hospital, Stockholm, Sweden
[7]ePAG & Chair SOD ITALIA - Italian Patients Organization for Septo Optic Dysplasia and other Neuroendocrine Conditions, Rome, Italy
[8]Aix-Marseille Université, Institut National de la Santé et de la Recherche Médicale (INSERM), U1251, Marseille Medical Genetics, Institut MarMaRa, Marseille, France
[9]EURORDIS - Rare Diseases Europe, Paris, France

Correspondence should be addressed to S M Webb: webbsusan4@gmail.com; swebb@santpau.cat

This paper forms part of a special series collated by European Reference Network on Rare Endocrine Conditions celebrating its fifth year. The guest editors for this section are Violeta Iotova, Jérôme Berherat, and George Mastorakos.

Abstract

Patient journeys are instruments developed by EURORDIS, The Voice of Rare Disease Patients in Europe, to collect patients' experiences; they may identify gaps and areas deserving improvement, as well as elements positively considered by affected persons. As with other patient-reported experiences, they can complete the clinical evaluation and management of a specific disease, improving the often long diagnostic delay, therapy, patient education and access to knowledgeable multidisciplinary teams. This review discusses the utility of such patient-reported experience measures and summarises the experiences of patients with acromegaly, Addison's disease and congenital adrenal hyperplasia from different European countries. Despite rare endocrine diseases being varied and presenting differently, feelings of not having been taken seriously by health professionals, family and friends was a common patient complaint. Empathy and a positive patient-centred environment tend to improve clinical practice by creating a trustworthy and understanding atmosphere, where individual patient needs are considered. Offering access to adequate patient information on their disease, treatments and outcome helps to adapt to living with a chronic disease and what to expect in the future, contemplating the impact of a disease on patients' everyday life, not only clin
social, financial, educational, family and leisure issues is desir
realistic expectancies for patients and can even lead to a redu
empowerment with patient-centred approaches to these com
should be contemplated more and more, not only for the ben
for the entire health system.

Key Words

- health perception
- patient journey
- Addison's disease
- acromegaly
- congenital adrenal hyperplasia

Stage of journey	Clinical presentation/symptoms	Patient/family needs	Ideal outcome/support (how to address the needs)
Pre-diagnosis: Diagnostic delay of 0.25–15 years before correct diagnosis First symptom or at diagnosis	Typical physical: headache, carpal tunnel sd, sweating, menstrual irregularity, feeling tired with less strength, growth of hands, feet & tongue (determining lisping), joint pains, facial changes perceived suddenly by third parties, appearance of more body hair, snoring or sleep apnea, weight gain, skin thickening, jaw growth with toothache, high blood pressure Feeling 'something is wrong' Psychological: insomnia, lack of concentration, less memory, unreasonable nervousness, depression, fits of rage, irritability Less frequently described: 'unexplained' very large placenta. Laryngeal ventricular hernia, change in smile	Two different scenarios: - No suspicion of disease, problems thought to be related to stress, ageing, weight gain or lifestyle changes - Years of repeated consultations with no recognition of the symptoms as apart of the disease, determining stress and impaired QoL Desire to understand what is going on and feel at peace Fear of reaction of relatives when disclosing the diagnosis High-quality information, care & empathy by specialised multidisciplinary reference centres to offer global care	Listening to the patient by all involved in their healthcare (primary care, specialists consulted, nurses) who complain they are not taken seriously despite knowing 'something is wrong'. Increase awareness of the disease at all health levels to reach an earlier diagnosis and prevent irreversible changes Empathy by primary care and prevent delays in referring patients or organising diagnostic tests Access to multidisciplinary expert teams not only for diagnosis but also for chronic follow-up Shorten waiting list for surgery, which is lived as very stressful.

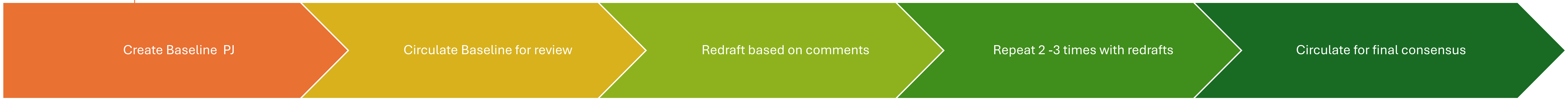
Lesson 3: While validation by lots of patients might not be possible in English, dissemination to all countries of English baseline yielded good engagement.

For Acromegaly

43 patients joined project

31 gave feedback

- ✓ Use EURORDIS template to structure data
- ❑ Data gathering via survey to fill gaps using Castor



Lesson 4: This process should be part of maintenance over longer time span to allow for updated versions to be published yearly/ every 2 year basis

3. Engage with Community

Using Delphi method:

- Refine the key details using content analysis
- Highlight areas with lacking support/ gaps
- Min. of 10 reviewers per redraft

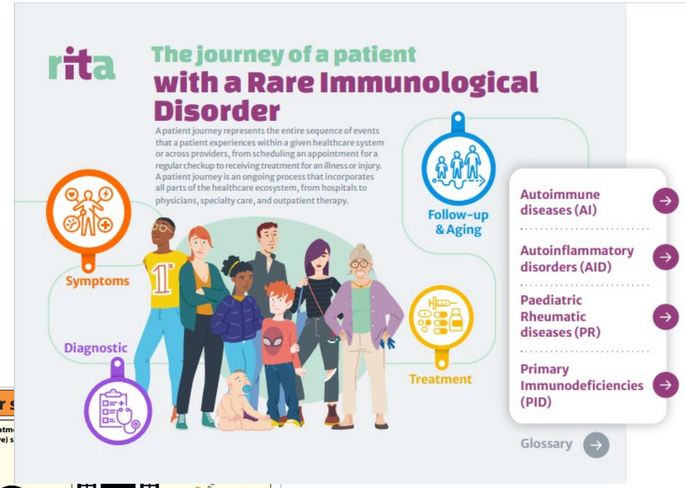
Create Baseline PJ → Circulate Baseline for review → Redraft based on comments → Repeat 2 -3 times with redrafts → Circulate for final consensus

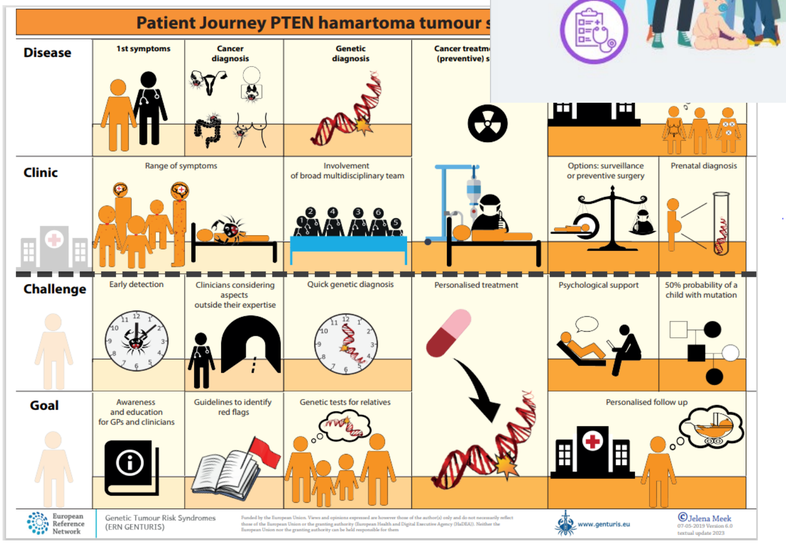
Due to start soon

4. Draft plan final Patient Journey

Discussions with MTG6 chairs & ePAGs on preferred format

Draft of data into graphical structure → PDF, handbook, video, webpage





ERN RITA ; ERN GENTURIS
Bolz-Johnson, Meek & Hoogerbrugge, 2020

5. Clinical review & Approval

In parallel with step 4, Clinical group of experts review final PJ version

- Via dedicated expert meeting
- At least 6 countries represented

Task:

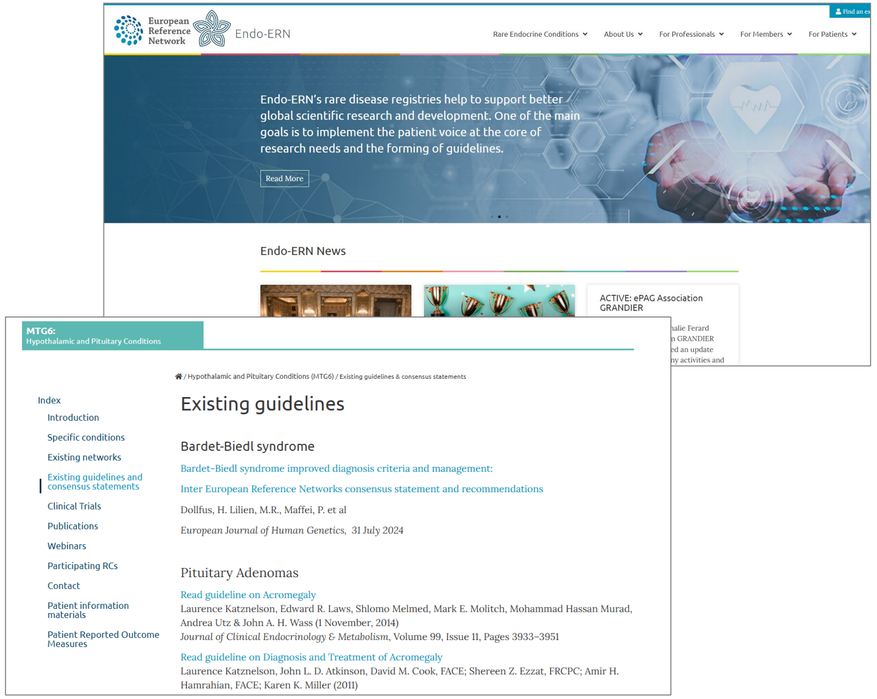
- Confirm medical data & clinical stage are accurate
- Check PJ is understandable

Lesson 5: Need input from ePAGs on most useful format this should take

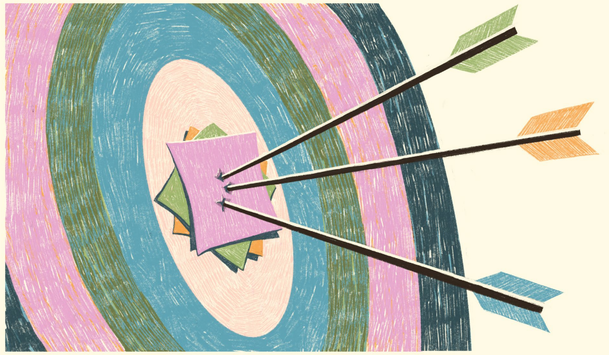
6. Put Journey into Action

Implementing PJ via Clinical Practise Guideline group in Endo-ERN.

1. Perform comparative review of the existing CPG within Endo-ERN - *Are all needs addressed?*
2. Create printable version of PJ for each member HCP
3. Publish to Endo-ERN website & disseminate to all Patient Advocacy Groups involved
4. Complete an Endo-ERN webinar detailing PJ



General Discussion



Our adaptions to our protocol will be added and the pilot with **Acromegaly** with continue, with a resulting version 2 patient journey created by end of 2025.

Additional lessons:

- Dedicated meeting with all MTG chairs prior to start of any patient journey is requested

- Mapping needed of existing rare endocrine patient journeys in process of being created e.g. Craniopharyngioma does not need a dedicated PJ as one is newly created

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Collaborative project on drug shortages

Johan de Graaf & Petra Breugmann

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Communication Update

Aimee Casey

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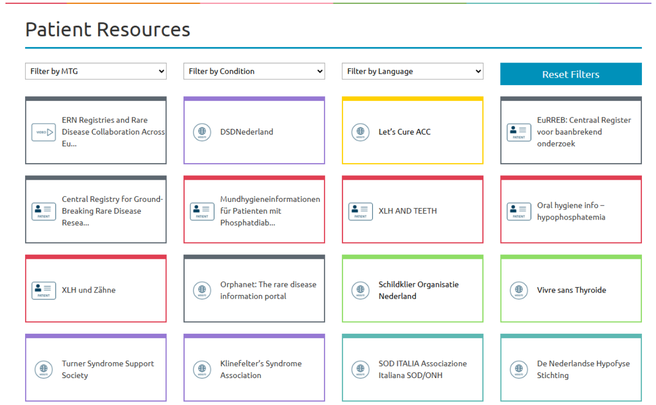
Patient Resources

- Over 100 searchable patient resources including links, documents and videos – corrections pending
- Compiling a list of other patient orgs for review by you as potential additions
- Are there resources that you find valuable?



Make changes to website: https://endo-ern.eu/website-updates/

Endo-ERN affiliation

- **Logo on your website**

- **New text**

The European Reference Network on Rare Endocrine Conditions (Endo-ERN) is a ***network of rare disease experts across Europe which can be accessed through your treating doctor.*** *Your doctor can request a virtual consultation with these specialists, bringing expert care to you, no travel required. Endo-ERN also signposts patient resources and you can contribute to a rare disease registry for research that will improve patient diagnosis and treatment. As an ePAG member organization, we recognize that the patient voice is an integral part of Endo-ERN and shapes the future of rare endocrine care across Europe.*

Webinar programme 2026

Potential topics – if we could plan 2 for 2026

- Holiday preparations – travelling with medication and emergency preparedness
- Working with rare disease
- Transition of care
- Working with your care team (as patient and carer)

- Any other ideas or if you are willing to speak please get in touch with me.
- Participation in MTG webinars as well

Odds & Ends

- ESE Adrenal Patient Forum 2025
- Thursday Eurordis will host an information webinar about the upcoming Rare Barometer – will share details afterwards

ePAG chairs: EURORDIS joint activities, other

Johan de Graaf & Petra Breugmann

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Important dates 2025 & 2026

- ePAGs & coordinators meeting, 02 December, online
- Endo-ERN General Assembly 2026, 20 and 21 April 2026, Athens, Greece
- ECE 2026, 9-12 May 2026, Prague, Czech Republic,
- ESPE 2026, 8-10 September 2026, Marseille, France