European Reference Network

Endo-ERN

Update RD committees ESPE and ESE

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Funded by
the European Union

Activities and future plans of the ESE Rare Disease Commitee

ESE & ESPE Joint Transition project - update

European Society of Endocrinology

ESE & ESPE Transition Project

A joint project between ESE & ESPE, on the topic of transition was proposed with the intention of completion/presentation at the joint ESE ESPE Conference in 2025

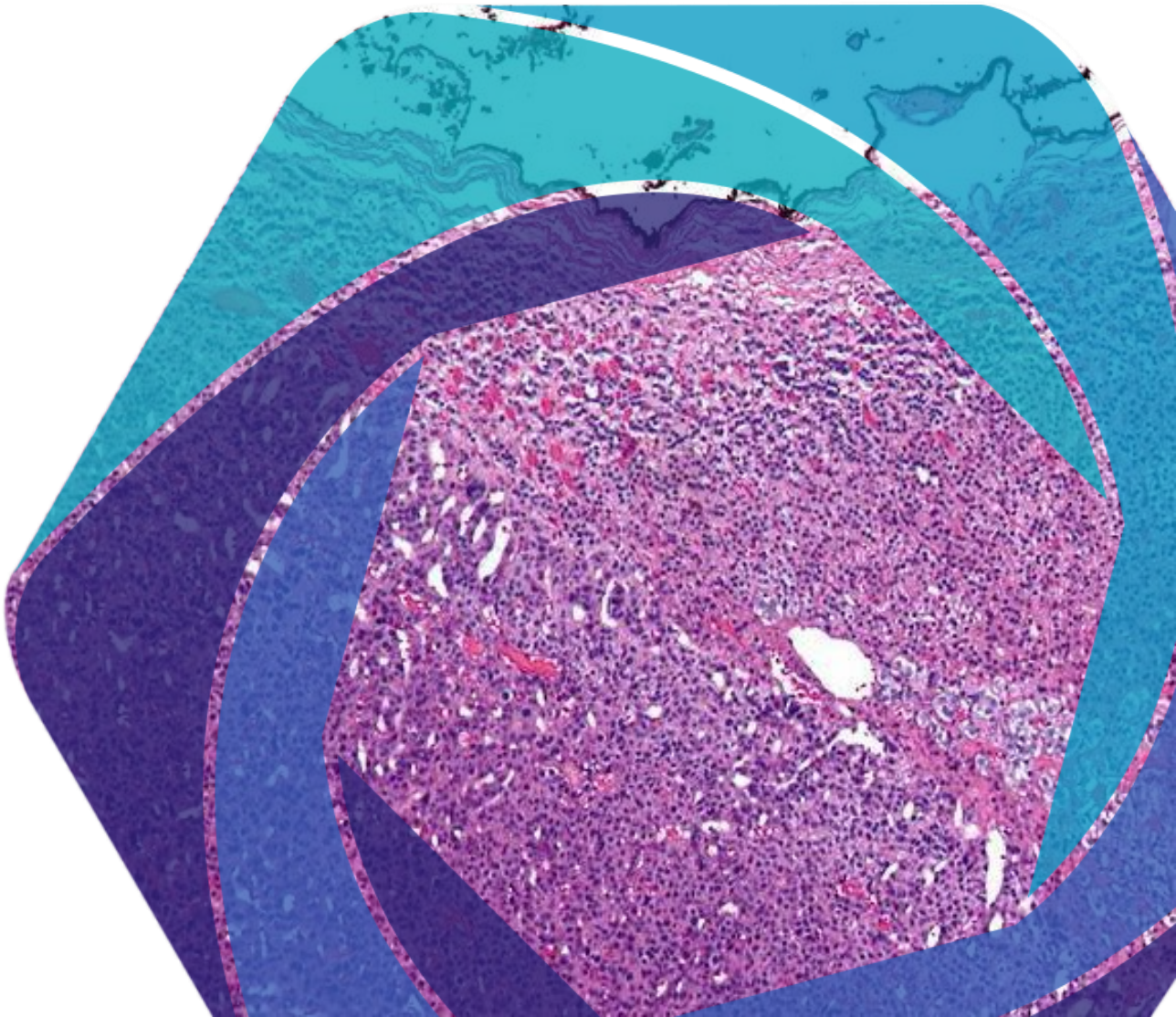
The objective of the group was to review existing literature on transition (in general, and on specific endocrine conditions) and make recommendations in the form of guidance for successful transition pathways.

An initial survey was completed to understand the perception of transition and existing transition pathways across Europe and different endocrine fields, after which background research into existing transition pathways was completed and reviewed. The methodology for systematic review was drafted, and a draft guidance statement has been produced.

ESE & ESPE Transition Project

- The draft guidance statement was developed through a process of review and discussion of the recommendations by focus groups of nurses, paediatric and adult endocrinologists.
- The draft document is in the process of being reviewed by a number of patient group representatives
- Submission to the European Journal of Endocrinology (EJE) and Hormone Research in Paediatrics (HRP), for co-publication, is expected in June 2025
- Presentation of the results at the congress will be on Tuesday 13th May at 15:35 – 17:05 CEST, by Sebastian Neggers and Enora Le Roux

Update on the Research Study/ESE Position Statement on Self-Management in Adrenal Insufficiency



Project aim

To understand patient needs for self-management support and reach consensus for patient education and support services in Europe

Self-management for patients with Adrenal Insufficiency (AI) involves complex behaviours across:

1) Optimising daily glucocorticoid (+ mineralocorticoid for PAI) replacement

2) Management of sick days and “steroid dosing”

3) Prevention and management of adrenal crisis.

Phase 1: Quantitative UK & international cross-sectional survey

Objective

To measure barriers and enablers to self-management in patients with AI (informed by Qualitative study)

Online survey

- Management of daily replacement, “sick days” and adrenal crisis;
- Measuring self-management behaviours and adherence to glucocorticoids (validated scales)
- Experience of patient education and care service provision

Recruitment: Patient Advocacy Groups and Endocrine Clinics (UK, Europe, Australia, USA/Canada)

UK Study completed Sept 2024

Three PAGs (N = 743) and 28 NHS endocrine centres (N = 1,180)

June – Dec 2025
PAGs from Europe, USA, Australia and 6-10 Endocrine Centres in Europe (pending R&D)

Questionnaire adaptations to each country and translations

Step 1:
Survey adaptations (Expert Panel of clinicians & PAGs), ≈100 collaborators from 15 countries

Step 2:
Translation and validation to each language (Expert Panel) & set up online survey in Qualtrics®

Step 3:
Dissemination of the survey via PAGs & Endocrine Centres in Europe

Timelines

PAGs: June – October 2025;
Endocrine Centres: June – Dec 2025 (to allow for R&D where needed)

Phase 2: Clinician survey, interviews & Delphi study

Stage 1
European online survey with endocrinologists & endocrine nurses (ESE, ECAS, EndoERN, SfE)

Stage 2
1:1 interviews to generate "*good practice*" vignettes across Europe (10-12 clinicians from S1

Stage 3
Generate & prioritise Position Statements (Nominal Group Technique with Study WG)

Stage 4
Delphi Study to test Position Statements: ≈50 clinicians & PAGs, 3-4 rounds for >75% consensus

Timelines

- Stages 1 & 2: June – Oct 2025 (conducted in parallel)
- Stage 3: Nov 2025 – Jan 2026
- Stage 4: Feb – July 2026

Ethics & Funding

- Ethics submitted in April 2025
- PT RF funded by City University & ADSHG: April-Nov 2025
- Grant application for £25K submitted to SfE on 7th May

Parallel project to the ESE Position Statement

Development and feasibility RCT of a web-based platform to support patients with Adrenal Insufficiency

Study Protocol: Llahana et al (2023).
Frontiers in Endocrinology

NIHR | National Institute for Health Research

Individualised
Interactive
Responsive

Potential for European funding and intervention implementation

Update on ESE Rare Calcium, Phosphate and Bone educational programme (ESE Rare-CaPaB)

European Society of Endocrinology

ESE Rare-CaPaB: ESE Rare Calcium, Phosphate and Bone disorders

The ESE Rare-CaPaB educational programme is a new expert led initiative to address the unmet needs of the calcium, phosphate and bone community. This programme will explore a range of rare calcium, phosphate and bone disorders with the following objectives:

- Identify the priorities to address concerning calcium, phosphate and bone disorders, and how to action these
- Understand and support the journey and management of patients, including in the transition from paediatric to adult care, as well as the ongoing needs of such patients throughout adulthood
- Establish ways to increase clinical awareness and diagnosis, including for the late onset forms
- Increase support and education of expert and non-expert healthcare audiences who regularly manage such patients

More details here: https://www.ese-hormones.org/education-and-training/educational-programmes/ese-rare-capab-an-ese-educational-programme-on-rare-calcium-phosphate-and-bone-disorders/

ESE Rare-CaPaB: ESE Rare Calcium, Phosphate and Bone disorders

The programme will focus on the following conditions:

- Osteogenesis imperfecta
- Hypoparathyroidism (with a focus on rare causes, i.e. genetic, autoimmune)
- Pseudohypoparathyroidism
- XLH and other rare causes of hypophosphatemia
- Hypophosphatasia

Overview of the ESE-Rare-CaPaB phases

The programme will be organised around 4 phases. Each phase is designed to inform the outcome of the next phase, some practical preparations for subsequent phases of work will happen in parallel. The 4 phases are:

Phase I – Scope and Prioritisation (6 months) - led by the steering committee and expert panel members. The final scope of the programme aims, objectives, final outputs and the measures of success will be established.

Phase II – Discovery and Development (6 months) – members of the steering committee supported by 10-20 experts, will establish agreement over the priorities identified in phase I.

Phase III – Synthesis and translation into Medical Education (9 months) – findings from phase II will be collated by the steering committee into a findings report, which will be sent for external review. These findings will be converted into a variety of medical education materials and activities to be disseminated to priority audiences.

Phase IV – Awareness, Communication and ESE integration (6 months) - delivery of programme of awareness and information communication campaigns on the programme conclusions to ESE members wider stakeholders. Consideration by ESE Committees, to integrate the outcomes into regular ESE educational activities.

Phase I: Scope and Prioritisation

✔ **European Expert Workshop on Rare Calcium and Bone Disorders (25-26 Oct 2024, Frankfurt)**

- Live workshop to setup a list of priority unmet needs

✔

- **Online survey**
  - Gain wider feedback from European stakeholders of the programme
  - Survey closed on Sunday 27 April 2025
  - Number of responses: **406**
  - Results currently under analysis

- **Virtual seminars: 11 & 18 June**
  - Broad audience of experts. To validate the planned work and gain further inputs, whilst offering opportunity to recruit future stakeholders

- **Discussion with patient advocacy groups**
  - To hear the practical challenges that patients are facing

Future 2025 Events

- **2 September 2025, 17:00-18:30 CEST – CAH; diagnostic and therapeutic challenges across the entire lifespan**

  Chair: Alberto Pereira

  - Presentation 1, 20 mins: Diagnostic and management challenges (with a focus on transition) - H Claahsen Van Der Grinten (NL)
  - Presentation 2, 20 mins: Novel therapeutic perspectives - H Falhammar (Sweden)
  - Presentation 3, 20 mins: Management of CAH in males (clinical case) - Riccardo Pofi (UK)
  - Q&A – 10-15 mins

Future 2025 Events

- **4 November 2025, 17:00-18:30 CEST – Recent therapeutic perspectives for rare endocrine diseases**

  Chair: Elena Valassi

  - Presentation 1, 20 mins: Long-acting rhGH: is it an improvement over daily rhGH? - M Maghnie (Genova, Italy)
  - Presentation 2, 20 mins: Palopegteriparatide : new perspectives for hypoparathyroid patients - L Rejnmark (Aarhus, Denmark)
  - Presentation 3, 20 mins – Novel therapies in acromegaly - Sabrina Chiloiro (Italy)
  - Q&A – 10-15 mins