ESE Rare Calcium Phosphate and Bone disorders educational programme (ESE Rare-CaPaB)

BRIEF SUMMARY REPORT – PHASE 1

October 2025

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ESE Rare Calcium, Phosphate and Bone disorders (Rare-CaPaB) – Introduction

The ESE Rare-CaPaB is an educational programme of the European Society of Endocrinology (ESE) designed to address the unmet needs of the calcium, phosphate and bone community, and to advance the knowledge and the medical education in five rare calcium, phosphate and bone conditions, and ultimately improve the awareness and the standards of medical care for patients suffering with these disorders. The aims of the ESE Rare-CaPaB are:

- 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 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 late onset forms
- Increase support and education of expert and non-expert healthcare audiences who regularly manage such patients

Why rare calcium, phosphate and bone disorders?

Rare disorders of calcium and phosphate metabolism, such as inherited causes of parathyroid hormone (PTH)-dependent or independent hypercalcemia, or hypoparathyroidism, inactivating PTH/PTHrp signalling disorders and X-linked and other genetic causes of hypophosphatemia, directly affect calcium and/or phosphate metabolism and alter the function of various target organs such as kidney and bone.

Metabolic bone diseases are caused by genetic abnormalities that may directly or indirectly affect the bone, or by other factors (hormones, tumours, diet, or drugs) that alter bone cell function. These diseases are rare, many are caused by changes in genes that can be passed on to children of affected adults; others may develop after birth from medical problems that were not inherited. For others, the cause remains unknown.

These disorders may result in abnormal growth and development of the skeleton and/or the inability to maintain healthy bones. Proper medical management of these rare conditions requires specialised training and experience. The issues faced with rare bone disorders, specifically, can be multi-fold, and may present quite an overlap with the general issues faced around other rare diseases due to:

- Insufficient awareness of the disease, leading to delays in diagnosis and treatment initiation
- Lack of research investment/options due to low patient numbers
- Complicated differential diagnosis requiring expert opinion
- Unfamiliarity with and/or limited access to disease reference and expert centres
- Availability and/or access to innovative drug treatments

A specific consideration for rare calcium, phosphate and bone disorders is that due to the onset of many disorders beginning in childhood, it is especially important to assess the best approaches to treating patients as they transition from adolescence to adulthood. In this regard, special attention needs to be considered regarding the:

- Structure of transitional care needed for patients that move from paediatric to adult settings
- Appropriate range of support and patient information provided to their family and caregivers
- Ongoing management of patients following transition from paediatric to adult care

What are the conditions that are being covered?

The ESE Rare-CaPaB is focused on the following five conditions:

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

Organisation & Governance

The ESE Rare-CaPaB governance structure includes two chairs and programme leads, a steering committee and an expert panel. These are further divided into five working groups, focused on each of the five identified conditions covered in the programme:

Co-Chairs and Programme Leads:

- Elena Tsourdi (Germany)
- Peter Kamenický (France)

Steering Committee:

- Giovanna Mantovani (Italy)
- Agnes Linglart (France)
- Carola Zillikens (Netherlands)
- Ralf Oheim (Germany)
- Andrea Palermo (Italy)
- Maria Yavropoulou (Greece)
- Outi Mäkitie (Finland)
- Corinna Grasemann (Germany)

Expert Panel:

- Liesbeth Winter (Netherlands)
- Adalbert Reimann (Austria)
- Natasha Appelman-Dijkstra (Netherlands)
- Luís Cardoso (Portugal)
- Tanja Sikjaer (Denmark)
- Martin Kuzma (Slovakia)
- Heide Siggelkow (Germany)
- Catherine Chaussain (France)
- Guiomar Perez de Nanclares (Spain)
- Neil Gittoes (UK)

- Justine Bacchetta (France)

Working Groups

Hypoparathyroidism:

- Heide Siggelkow (group leader)
- Andrea Palermo
- Luís Cardoso
- Liesbeth Winter

Pseudohypoparathyroidism:

- Giovanna Mantovani (group leader)
- Tanja Sikjaer
- Guiomar Perez de Nanclares
- Agnès Lignart
- Corinna Grasemann

Osteogenesis imperfecta:

- Neil Gittoes (group leader)
- Outi Mäkitie
- Ralf Oheim

XLH / Hypophosphatemia:

- Catherine Chaussain (group leader)
- Natasha Appelman-Dijkstra
- Adalbert Raimann
- Justine Bacchetta

Hypophosphatasia:

- Carola Zillikens (group leader)
- Martin Kuzma
- Maria Yavropoulou

Target Audience & Beneficiaries

The programme is designed to be of benefit to various clinical communities:

- International parathyroid disorder experts
- General endocrinologists
- All ESE Members
- Non-endocrine practitioners
- Patient stakeholder groups

Sponsors & Supporters

The ESE is grateful to Alexion, Ascendis Pharma, BridgeBio, Inozyme Pharma and Kyowa Kirin for their support to this programme via restricted educational grants.

None of the supporters played any role in the objectives, aims, scope, faculty selection, nor will participate in the development and outputs of the educational programme. All steering committee and expert panel members declared any conflicts of interest in respect of these organisations and received ESE approval to take part.

Programme website

A webpage for this educational programme has been created on the ESE website. More details regarding ESE Rare-CaPaB can be found in the webpage: 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: Phases & Activities

The ESE Rare-CaPaB educational programme is organised around 4 initial phases of activity. Each phase is designed to inform the outcome of the next phase of activity, some practical preparations for subsequent phases of work will happen in parallel. The four phases are:

Phase I – Scope and Prioritisation: 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: members of the steering committee supported by 10-20 community experts, will establish agreement over the priority objectives identified in phase I. The steering committee will organise the programme which will include presentations and discussion on the key topics and disorders.

Phase III – Synthesis and translation into Medical Education: findings from phase II will be collated by the steering committee into a final 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: delivery of programme of awareness and information communication campaigns on the programme conclusions to ESE members and wider stakeholders. Consideration by ESE Committees, to integrate the outcomes into regular ESE educational activities.

Below, we summarise the key activities and initiatives within each phase. The Phase 1 is currently completed, and in the next pages of this report, a detailed overview of each activity will be provided.

Phase I – Scope and Prioritisation

- European Expert Workshop on Rare Calcium and Bone Disorders
  - held on 25-26 October 2024, in Frankfurt, Germany

- Online survey
  - Aimed to gain wider feedback from European stakeholders of the programme
  - Survey closed on Sunday 27 April 2025. Number of responses: 407.
  - Results were analysed and discussed in the Virtual Expert Seminars and PAG Webinar

- Virtual Expert Seminars
  - Broad audience of experts in field
  - To validate the planned work and gain further inputs, whilst offering opportunity to recruit future stakeholders
  - SEMINAR 1: Wednesday, 11 June 2025, 15:00 – 17:30 CEST
  - SEMINAR 2: Wednesday, 18 June 2025, 15:00 – 17:00 CEST

- Patient advocacy groups (PAG) Webinar
  - To hear the practical challenges that patients are facing
  - PAG WEBINAR: Monday, 6 October 2025, 14:00 – 17:00 CEST

Phase II – Discovery and Development

- Clinical working groups
  - To form consensus on the priority unmet needs questions as identified in phase 1
  - To create summary findings report with objectives and joint opinions
- Meeting of the steering committee with the clinical working groups
  - To ensure cohesion of the findings across the different working groups and encourage further collaboration

Phase III – Synthesis and translation into Medical Education

- Session/meeting at European Congress of Endocrinology
- Creation of medical education materials and activities
- Awareness-raising activities
- Findings report of key consensuses established by the programme

Phase IV – Awareness, Communication and ESE integration

- Awareness campaign around the educational materials and programme outputs
- Integration of the outcomes into regular ESE educational activities

ESE Rare-CaPaB: Phase I

The ESE Rare-CaPaB Phase I is now completed. The formation of the steering committee and expert panel was undertaken during the preliminary stages of the programme. The co-chairs of Rare-CaPaB (Elena Tsourdi and Peter Kamenický) were proactive in forming a steering committee and expert panel after combining their own suggestions with the recommendations from the following ESE Committees, Groups and stakeholders:

- ESE Clinical Committee
- ESE Rare Disease Committee
- ESE Education Committee
- Focus Area of Calcium and Bone
- European Society for Paediatric Endocrinology (ESPE)
- European Registries of Rare Diseases (EuRRECa)
- European Calcified Tissue Society (ECTS)

The steering committee and expert panel consist of a comprehensive, inclusive, diverse and equitable mix of European experts, to represent different geographical areas in Europe and to ensure representation of the different unmet needs and challenges across distinct countries.

European Expert Workshop on Rare Calcium and Bone Disorders

The European Expert Workshop on Rare Calcium and Bone Disorders was held on 25-26 October 2024, in Frankfurt, Germany. This kick-off meeting took place after the formation of the Steering Committee and Expert Panel, where the experts had the first opportunity to meet, discuss and harness expert opinions while setting up the foundations and key steps/milestones of ESE Rare-CaPaB, including the initial drafting of the unmet needs list, and the brainstorming around the scope, aims and format of the envisaged online survey. The meeting was very successful, with excellent attendance and engagement from key experts ensuring key discussions took place.

Survey - Refining and prioritising unmet needs in rare calcium, phosphate and bone disorders

An online survey was created by the ESE Rare-CaPaB Steering Committee and Expert Panel to gain broader feedback from a wide range of clinicians and other healthcare professionals, as well as from European stakeholders regarding the key educational areas of the ESE Rare-CaPaB programme to be explored in relation to the five conditions covered in the programme.

The development of the survey commenced at the In-Person Workshop in Frankfurt in October 2024, and it was further constructed over a few online meetings and thanks to the work of each respective working group. The main focus was to create a list of 7 items where the working groups experts felt there were needs and areas of uncertainty, controversy or lack of knowledge, thus requiring special attention in terms of medical education and clinical awareness.

The survey was sent to all ESE members, as well as to wider audiences in collaboration with stakeholder organisation partners, such as Endo-ERN, ESPE, ECTS, and national endocrine societies. The survey closed on 27 April 2025, and received a total of **407 responses** from 67 different countries.

The survey was mainly completed by clinicians and clinician scientists, but a few other healthcare professionals also provided replies to the survey, such as nurses and dentists. The overwhelming majority of the respondents were endocrinologists (81.7%), followed by paediatric endocrinologists (21.1%). Seventy-three percent of the respondents were members of the ESE.

The results were first discussed by the ESE Rare-CaPaB Steering Committee and Expert Panel during an in-person meeting that was held on 12 May 2025 during the Joint Congress of ESPE and ESE in Copenhagen, Denmark. The results allowed the Steering Committee and Expert Panel of ESE Rare-CaPaB to have preliminary discussions in terms of which areas to focus on within the programme, and which unmet needs and knowledge gaps might be covered in the future educational materials and outputs deriving from the programme.

The results of the survey were then reviewed and discussed during the two Virtual Expert Seminars in June 2025, where external invited experts in the field of bone, calcium and phosphate disorders were able to comment and share their views. Later in October 2025, the results of the survey were further presented to an audience of patient advocacy group representatives during a 3-hour webinar, where participants were able to comment and express their opinions on the results.

Ultimately, the results from the survey will help in prioritising the key educational areas to be covered during the ESE Rare-CaPaB educational programme, and will help in guiding the creation of the educational materials and outputs of the programme.

Virtual Expert Seminars

After the steering committee and expert panel of ESE Rare-CaPaB drafted a proposed set of priority unmet needs which were incorporated in the online survey, and after having the answers to the online survey, the next stage of the Rare-CaPaB programme, consisting of two virtual expert seminars, commenced.

For these online events, a broad range of experts in bone, calcium and phosphate disorders were identified and invited by the Steering Committee and the Expert Panel of the Rare-CaPaB. The main objectives were to validate the planned work, gain further inputs, and finetune the unmet needs and educational priorities of the programme, as well as to discuss outstanding questions, whilst offering an opportunity to recruit future stakeholders.

The expert virtual seminars were held on 11 and 18 June 2025. The first seminar covered hypoparathyroidism, pseudohypoparathyroidism, and osteogenesis imperfecta, while the second seminar was focused on XLH and hypophosphatasia.

During these virtual expert webinars, it was discussed various aspects of the five conditions covered in the Rare-CaPaB programme, which included discussions on the survey results, educational priorities and upcoming tasks/events, which are ultimately focused on developing tailored educational materials and improving patient care. It was also emphasised the need and importance of collaboration with patient advocacy groups and addressing challenges of patients transitioning from paediatric to adult care.

As key conclusions and next steps to be taken after these expert seminars, it was highlighted the need to proceed with the next programme steps, which are the creation of a proposal for a Symposium at ECE 2026 and to organise further the PAG webinar. After developing these 2 key activities, it was also decided that the programme will further advance to its phase 2 and 3 aimed to develop and create medical education materials and outputs such as a Symposium at ECE 2026, patient leaflets, summary finding reports, clinical vignettes/medical case questions, and an ESE library course.

PAG Webinar

A Patient advocacy groups (PAG) Webinar took place online on Monday, 6 October 2025, 14:00 – 17:00 CEST. A wide range of PAG associations focused on the five conditions covered in the ESE Rare-CaPaB programme were identified, and formal invitations were sent for their participation. The aim of this webinar was to understand the challenges that patients are facing and identify unmet needs.

The webinar was attended by 18 representatives from a total of 11 PAG associations:

- Asociación Española de Pseudohipoparatiroidismo (PHP) – AEPHP
- Asociación Nacional Huesos de Cristal OI España, AHUCE
- Brittle Bone Society UK
- GACI Global
- Hifo-der
- HypoPT Eastern Europe Group
- International XLH Alliance (IXLHA)
- Netzwerk Hypopara

- Osteogenesis Imperfecta Federation Europe (OIFE)
- Phosphatdiabetes EV
- Rare Diseases Greece (RDG)
- XLH Denmark

The meeting covered various aspects of hypoparathyroidism, pseudohypoparathyroidism, osteogenesis imperfecta, XLH and other rare causes of hypophosphataemia, and hypophosphatasia, including a presentation on the survey results, followed by a dynamic discussion with various representatives of PAGs from across Europe. Overall, the importance of collaboration with PAGs in addressing the unmet needs of patients with such conditions across Europe in many dimensions was emphasised, from diagnosis, management, access to treatments and tertiary centres, reimbursements of medications, coordination of care, quality of life, complexities of defining severity, and other topics.

During the PAG Webinar, some transversal matters across the 5 conditions were discussed. The group discussed healthcare access and reimbursement for rare bone diseases. From the discussion, it emerged that there is a significant variation in patient support across countries, with some countries having well-established PAG associations while others lack organised advocacy groups. It was further emphasised the importance of coordination among stakeholders and proposed taking responsibility for cross-cutting issues like pain and fatigue, while concerns were also raised about the limitations and lack of good quality of life measurements at present, and the need for better data collection.

Next steps – moving forward to Phase II and III

As the programme moves forward to Phases II and III, with the ultimate aim of developing and creating the educational materials and outputs based on the insights and input received from the various initiatives of Phase I, the ESE Rare-CaPaB Steering Committee and Expert Panel have already delineated and consolidated a few initiatives and activities described below.

Symposium at ECE 2026 (Prague, Czech Republic)

A symposium proposal for the ECE 2026 has been drafted by the Rare-CaPaB Steering Committee and Expert Panel and submitted to the Programme Organising Committee of ECE 2026. The symposium proposal was accepted, and a Rare-CaPaB symposium is now part of the main congress programme.

Online courses for the ESE Library

ESE has recently developed and launched the Learning Management System (LMS) project. The LMS is part of a larger digital infrastructure project, and it includes the possibility to add recorded sessions and online courses that can be seen live or on-demand by ESE members. As part of the LMS, the ESE has created an ESE Online Library, where such content can be placed. The Rare-CaPaB Steering Committee and Expert Panel will devise an online course covering each one of the 5 conditions. This will be a great opportunity for ESE members, including endocrinologists and in-training residents, which will improve the skills, cover the knowledge gaps and increase awareness for rare calcium, phosphate and bone disorders.

Written educational materials: manuscripts for a wide audience of healthcare professionals, patient leaflets and other educational materials

A key initiative of the ESE Rare-CaPaB educational programme is producing long-lasting educational materials and written information to be available to a wide public. Some of these materials will include review and opinion manuscripts to be published in high-impact and widely read journals (e.g. the European Journal of Endocrinology), patient leaflets covering areas of topics that are currently lacking, and potentially other educational materials (e.g. clinical vignettes, exam questions, etc)