Clinical decision-making in rare bone diseases: a European survey among EPOS and ERN-BOND members

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Bakgrunn

Rare bone diseases (RBDs) comprise over 700 heterogeneous disorders with low prevalence, diagnostic delays, and complex multidisciplinary needs. Despite progress, challenges persist, including inconsistent diagnostic pathways, lack of standardized decision-making frameworks, and variability in interdisciplinary collaboration. Many clinicians encounter too few cases to build experience, and guidelines are often incomplete. To address this, ERN-BOND (European Reference Network for Rare Bone Disorders) and the European Paediatric Orthopaedic Society (EPOS) initiated a survey to map current practices and identify unmet needs in RBD care across Europe.

The primary aim is to describe current clinical decision-making in RBDs among EPOS members and outline a minimum standard for patient assessment. Secondary aims are to identify available tools and resources, explore interdisciplinary collaboration, examine approaches to shared decision-making, and assess educational needs.

Materiale og metoder

An anonymous online survey (Google Forms) was distributed to EPOS and ERN BOND members, targeting clinicians, researchers, and patient representatives involved in RBD care. The survey includes 35 questions (multiple-choice, Likert scale, and open text). Topics cover diagnostic work-up, treatment decision-making, interdisciplinary care, use of gait analysis, transition from paediatric to adult care, and perspectives on future needs. Responses will be collected with reminders sent to optimize participation. Data will be analysed descriptively for quantitative items and thematically for free-text responses.

Resultater

At the time of abstract submission, data collection is ongoing. Findings will describe diagnostic and therapeutic practices, highlight variations in interdisciplinary care, and provide insights into barriers, facilitators, and educational needs. Results will be available and be presented at the October 2025 meeting.

Konklusjon

This is the first pan-European survey within ERN-BOND and EPOS to systematically capture clinical decision-making in RBDs. The results will inform guideline development, decision-support tools, and educational resources, ultimately contributing to harmonized care for children and adults with rare bone diseases.