

ERN BOND WP4 MADRID MEETING

Meeting short report

Location: Hospital Universitario La Paz in Madrid, Spain

Date: 3rd March 2025

1. REPORT

ERN BOND WP4 MADRID MEETING (3rd March 2025)

On Monday, March 3rd, the ERN BOND WP4 meeting took place at Hospital Universitario La Paz in Madrid, Spain. Ten ERN BOND members from seven Healthcare Providers (HCPs) across Spain, Italy, Portugal, Denmark and the Netherlands participated. Eight participants attended in person and two attended online.

The meeting focused on one of the WP4 objectives: improving genetic testing for rare bone diseases (RBDs).

This meeting covered the discussion of common errors and pitfalls observed in clinical practice and research, genomic and clinical complexity of skeletal dysplasias and good practice suggestions and guidelines for genetic testing.

Participants worked on structuring an article that will address these key challenges, with the aim of improving genetic testing in RBDs, and subsequently improving patient care.

The group also discussed the proposal for a European Molecular Quality Assurance Network (EMQN) skeletal dysplasia scheme. EMQN provides quality assurance tools and knowledge to the human genomic testing community through External Quality Assessment (EQA) schemes. More information about EMQN is available at <https://www.emqn.org/>.

As an outcome of the meeting, a work plan was established with tasks assigned to each participant. In the coming months, the group will continue meeting online to monitor progress towards achieving this WP4 objective.

This meeting represents another step forward in the effort to improve the diagnosis and care of patients with rare bone diseases across Europe.

List of the participants in the working group attending to this meeting:

NAME	AFFILIATION
Karen Heath	Hospital Universitario la Paz, Madrid, Spain (ES15)
Silvia Modamio	Hospital Universitario la Paz, Madrid, Spain (ES15)
Miguel Barroso	Hospital Universitario la Paz, Madrid, Spain (ES15)
Monique Losekoot	Leiden University Medical Centre, LUMC, Leiden, the Netherlands (NL05)
André Travessa	Centro Hospitalar Universitário Lisboa Norte (CHULN), EPE, Lisboa, Portugal (PT05)
Alice Moroni	Istituto Ortopedico Rizzoli, Bologna, Italy (IT51)
Elena Pedrini	Istituto Ortopedico Rizzoli, Bologna, Italy (IT51)
Trine Østergaard Nielsen	Aarhus Universitets Hospital, Aarhus, Denmark (DK01)
Sérgio Sousa (online)	Centro Hospitalar e Universitário de Coimbra, EPE, Coimbra, Portugal (PT03)
Maria Luisa Brandi (online)	University Vita-Salute San Raffaele, Milan, Italy

8th Hybrid Meeting of the Hospital Universitario la Paz Skeletal dysplasia Multidisciplinary Unit (UMDE-ERN BOND), Madrid (18th March 2025)

On Tuesday, March 18th, it occurred the 8th Hybrid Meeting of the Hospital Universitario la Paz Skeletal dysplasia Multidisciplinary Unit (UMDE-ERN BOND) in Madrid, Spain.

This event brought together 268 participants from seven different countries: Netherlands, France, Argentina, Portugal, USA, Chile and Spain. This event addressed the clinical and molecular diagnosis of skeletal dysplasias: from research to clinical practice.

The meeting consisted in three sessions, including the discussion of clinical cases of patients with skeletal dysplasias

The first session focussed on Spondylo-epi-metaphyseal Dysplasias (SEMD), in which Karen Heath, co-leader of ERN BOND WP4, co-presented the session's main talk on clinical, radiological, and genetic approach of SEMD. She was also a moderator for the rest of the sessions.

The second session focussed on other skeletal dysplasias. This session included a talk of Miguel Barroso, one of the project managers of ERN BOND WP4, on ERN BOND's activities for the coming years, with a focus on the objectives of ERN BOND WP4. This talk described the recent ERN BOND WP4 actions towards the objectives of improving genetic testing for rare bone diseases and improving healthcare in pregnancies and the use of the platform Clinical Patient Management System 2.0 (CPMS 2.0), as well as future collaborative efforts to improve care for rare bone diseases.

The third and final session was dedicated to the presentation of clinical cases from different experts, promoting the interactive exchange of knowledge and experiences among specialists from multiple disciplines. It was emphasized the importance of multidisciplinary collaboration between experts and the integration of genetic, clinical and radiological findings to improve diagnosis and treatment of patients with skeletal dysplasias.



https://ec.europa.eu/health/ern_en



<http://ernbond.eu/>



Funded by the
European Union