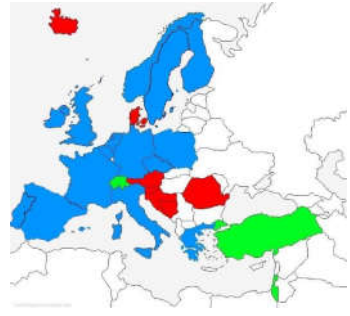




ERN-BOND Surveys on Diagnosis of Osteogenesis Imperfecta: the healthcare professional and patients experience

Matias de la Calle (1), Marina Mordenti (1), Manila Boarini (1), Maria Gnoli (1), Luca Sangiorgi (1)

(1) Medical Genetic Department, IRCCS Istituto Ortopedico Rizzoli



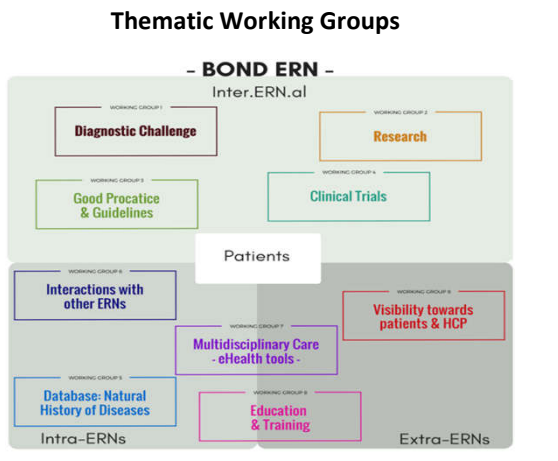
ERN-BOND is the European Reference Network for Rare Bone Disorders.
It brings together 38 highly specialised healthcare providers from 10 EU Member States.

- To foster holistic, multidisciplinary and patient-centred care
- To connect the best healthcare professionals with the best researchers, in order to improve access to cross-border medical expertise in line with Directive 2011/24/EU3.

The network has selected one of the most common rare bone diseases, osteogenesis imperfecta, as an area of focus, to understand the common challenges in diagnosing rare bone diseases and to provide recommendations for improving referrals, reducing diagnostic errors and shortening diagnostic delays.

THE ERN-BOND WHITE PAPER ON DIAGNOSING OSTEOPOROSIS IMPERFECTA

- Overview of the current situation relating to diagnosing *osteogenesis imperfecta*
- Identify the key challenges and potential solutions to further reduce these shortcomings, and improve the patient experience.



Results

THE SAMPLE: QUESTIONNAIRE FOR HCPS

Figure 2: HCPS questionnaire – distribution of respondents among participating countries

Figure 3: HCPS questionnaire – area of expertise of the respondents

DIAGNOSTIC PATHWAY

Figure 7: HCPS questionnaire – estimated time to confirm diagnosis (in years)

Figure 8: Patients questionnaire – average time from referral to diagnosis (in years)

THE SAMPLE: QUESTIONNAIRE FOR PATIENTS

Figure 4: Patients questionnaire – geographical distribution of the respondents

WRONG DIAGNOSIS

Figure 10: Patients questionnaire – patients receiving a wrong diagnosis

Figure 11: HCPS questionnaire – patients with a wrong diagnosis

Conclusion

BARRIERS

- Lack of information regarding rare bone disorders among both health doctors and general specialists
- Lack of awareness of all doctors at general public
- Difficulties of accessing specialist care
- Reluctance in performing the genetic test
- Shortage of staff allows in the Emergency Room

SUCCESS FACTORS

- With an early referral to the CR specialist, the patient's overall experience and quality of life are supported by the more positive from the moment in the diagnostic response, even if this occurs during pregnancy.
- Direct access to an CR specialist, guarantee faster engagement and follow-up.
- When possible, linkage with local or virtual patient groups provides a supportive platform.
- When Excellence Centres are easily accessible, most across borders, the overall patient journey is easier, shorter and altogether more efficient.

POTENTIAL SOLUTIONS

- There is a clear need to continue with education and training, involving professionals and CRN doctors, both general and specialist, to improve the awareness of the importance of having CRN specialists, especially among family doctors and emergency department healthcare professionals.
- Continuing professional education for the multi-professional team will also be essential to ensure the best patient experience.
- There are high investments in diagnosis, prevention and follow-up, the majority of HCPS acknowledge that during their clinical practice, guidelines for diagnosing CRN bone disorders are essential and address existing legislation.
- An CRN in genetic disease, performing a genetic test can help confirm the presence of the disease in the early stages of life. This would contribute towards preventing any other diagnosis, and preventing a further quality of life positive with care and diagnosis.



For information:
matiasignacio.delacalle@ior.it