

An ERN-BOND White Paper On Diagnosis

Luca Sangiorgi, ERN-BOND Coordinator, Istituto
Ortopedico Rizzoli, Italy

Meena Balasubramanian, Valerie Cormier-Daire,
WG "Diagnostic Challenge", ERN BOND

Inês Alves, ERN BOND EPAG, ALPE Foundation



ERN-BOND White Paper on Diagnosis of Osteogenesis Imperfecta: Synopsis



ERN-BOND



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 OSTEOGENESIS IMPERFECTA

The objective of the White Paper is to provide an overview of the current situation relating to diagnosing *osteogenesis imperfecta* in the 10 Member States represented within ERN-BOND.

As collective experience points towards delays in detection, this paper's main aim is to identify the **key challenges and potential solutions to further reduce these shortcomings, and improve the patient experience.**



THE SAMPLE: QUESTIONNAIRE FOR HCPS

- **Incidence:** The majority of HCPs indicated that they were unaware of any incidence of OI in their country.
- **Prevalence:** When it comes to prevalence, respondents provided an accurate estimate (ca 0.9 in 10,000), which is slightly lower than the average European estimates (1-5 in 10,000).
- However, more than 60% of HCPs **suspected** the presence of OI in undiagnosed patients in their Excellence Centre. 61% of the HCPs also indicated that they **confirmed the OI diagnosis** in more than 50% of the cases referred.

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

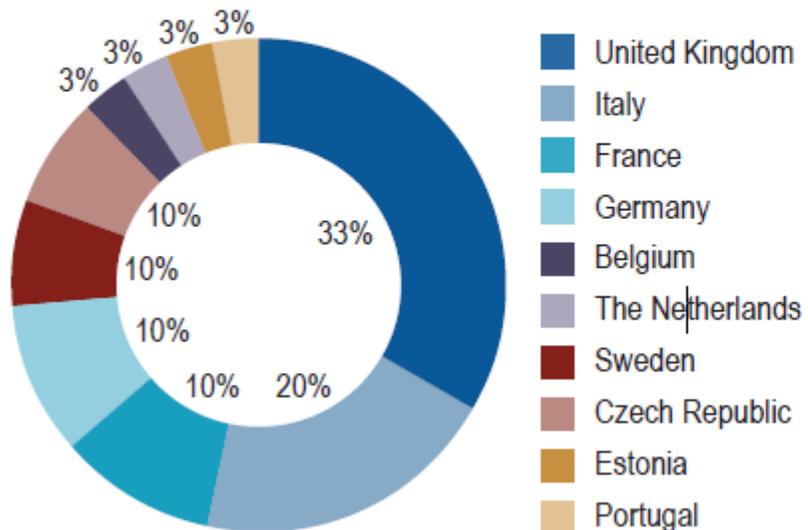
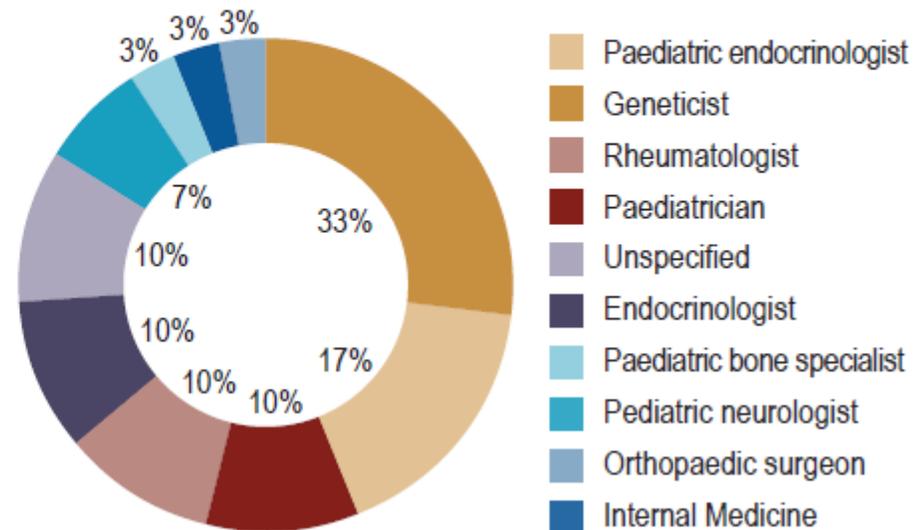


Figure 3: HCPs questionnaire – area of expertise of the respondents



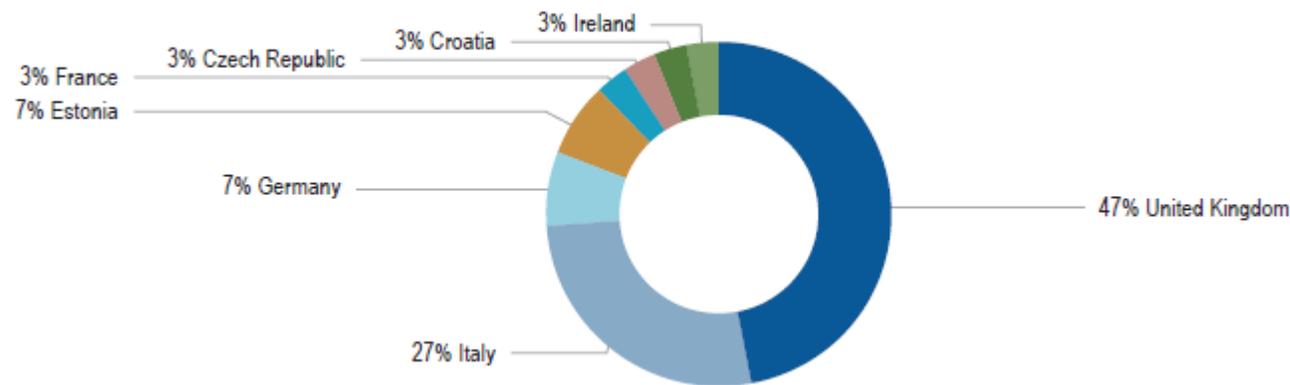


THE SAMPLE: QUESTIONNAIRE FOR PATIENTS

The age of the OI patients surveyed either directly or indirectly (via carers) varied greatly, ranging from new-borns to 71 years old.

The majority of respondents (63%) said that they were not involved in a **patient organisation**. None of the carers indicated that they were members of an OI organisation.

Figure 4: Patients questionnaire – geographical distribution of the respondents





DIAGNOSTIC TESTS

- Genetic test & family history:
 - 60% of patients reported that a **genetic test** was performed to confirm the diagnosis;
 - On the HCPs side, almost half of the specialists stated that **genetic testing had contributed to accelerating diagnosis**.
- The questionnaire for HCPs also showed a **high level of heterogeneity in the diagnostic and testing procedures** used by the different centres across the various countries.

Figure 5: HCPs questionnaire – family history of OI

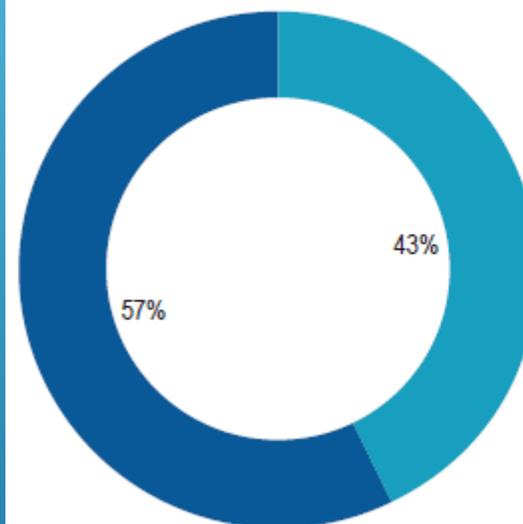
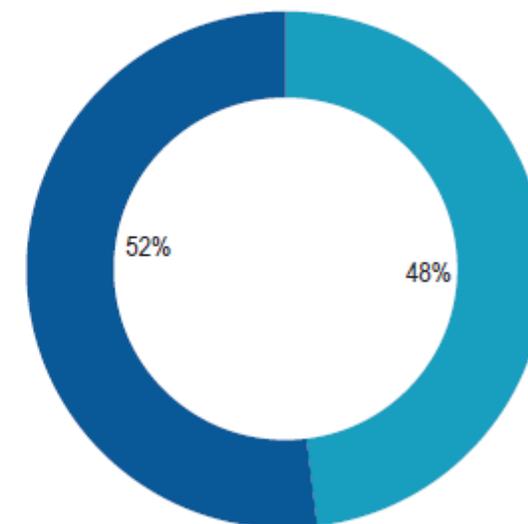


Figure 6: Patients questionnaire – family history of OI



 No  Yes

DIAGNOSTIC PATHWAY



European Reference Network

for rare or low prevalence complex diseases

Network Bone Disorders (ERN BOND)

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

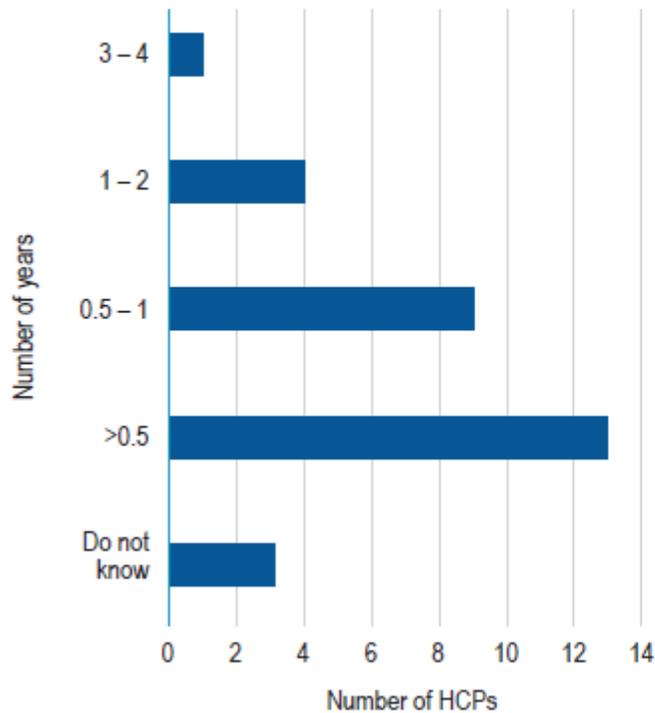
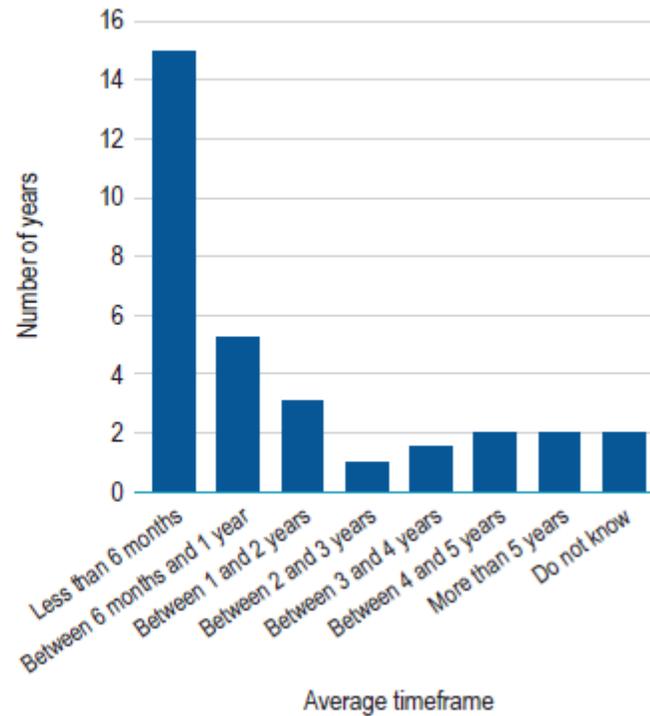


Figure 8: Patients questionnaire – average timeframe to receive diagnosis (in years)



From the time of the disease's first symptoms emerging to obtaining an accurate diagnosis, the majority of both HCPs and patients indicated an average period of **less than 6 months to confirm the diagnosis**. This corresponds to **less than 5 visits between primary and specialised care**.

This denotes that, within this sample, **the time taken for diagnosing OI is not as long as for other rare diseases**.

However, despite the 6-month average, a fifth of overall respondents indicated that the procedure could take up to 4 years.

WRONG DIAGNOSIS

Figure 10: Patients questionnaire – patients receiving a wrong diagnosis

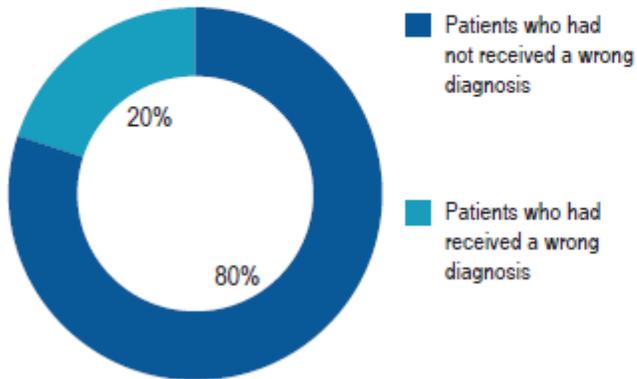
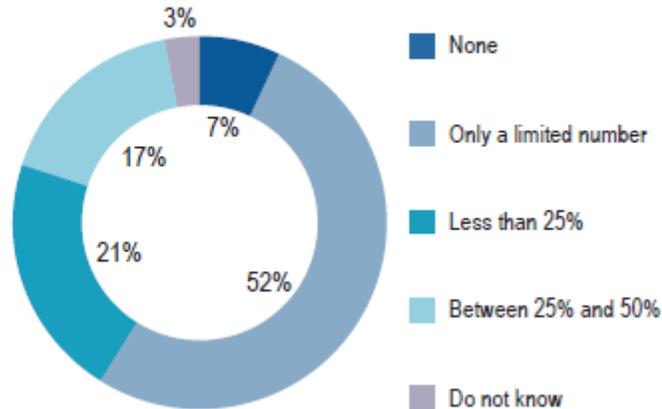


Figure 11: HCPs questionnaire – patients with a wrong diagnosis



When patients were asked whether they had ever received a **wrong diagnosis**, 20% of them said that this was initially the case.

The majority of HCPs estimated that only a limited number of their patients had received a wrong diagnosis before being referred to their Excellence Centre.

Yet HCPs identified **osteoporosis** and **child abuse** and as the most common reasons for a misdiagnosis.



**European
Reference
Network**

for rare or low prevalence
complex diseases

 **Network**
Bone Disorders (ERN BOND)

BARRIERS



Lack of information regarding rare bone diseases among both family doctors and medical specialists.



Lack of awareness of OI among the general public.



Difficulties in accessing specialised care.



Reluctance in performing the genetic test.



Suspicion of child abuse in the Emergency Room.



SUCCESS FACTORS



With an early referral to the OI specialist, the patient's overall experience and quality of life are reported to be more positive from the moment the disease is suspected, even if this occurs during pregnancy.



Direct access to an OI specialist guarantees better management and follow-up.



When possible, linkage with local or virtual patient groups provides a supportive platform.



When Excellence Centres are easily accessible, even across borders, the overall patient journey is easier, shorter and altogether more efficient.



European
Reference
Network

for rare or low prevalence
complex diseases

 Network
Bone Disorders (ERN BOND)

POTENTIAL SOLUTIONS

The data collected with this survey reveals that both physicians and patients agree on a need for **rapid referral to the specialised centre**, so as to ensure a **timely diagnosis of OI** and to improve care.



Since OI is often confused with other, more common diseases, including osteoporosis and child abuse, both groups underlined the importance of raising OI's awareness, especially among family doctors and emergency department healthcare professionals.



Continuous professional education for these groups, as well as for the members of the multi-disciplinary team was also identified as essential for avoiding delays in diagnosis.



Given the high heterogeneity in diagnostic procedures and testing, the majority of HCPs acknowledged that devising 'best clinical practice guidelines' for diagnosing OI could help standardise processes and address existing inequalities.



As OI is a genetic disease, performing a genetic test can help confirm the presence of the disease in the early stages of life. This would contribute towards ensuring an early diagnosis, and providing a better quality of life for patients with rare bone diseases.

CONCLUSIONS AND POLICY RECOMMENDATIONS



Awareness-raising activities of rare bone diseases among primary and emergency care practitioners, through training to identify the signs and symptoms of rare bone disorders for healthcare professionals, family doctors and emergency care practitioners, in order to improve referrals to the reference centres.



Supporting the creation of national clinical networks connected to the Excellence Centres in order to provide accurate diagnosis and clinical support for patients and families.



Developing European guidelines for OI diagnosis to facilitate rapid and accurate diagnosis through standardised procedures, and to reduce the differences between centres and countries.



Empowering patients and their carers through strategies to support the development of local support groups to provide them with high quality information.

The survey identifies areas for improvement that need a **multi-stakeholder approach** to increase standards and accelerate OI diagnosis, as well as accelerate the diagnosis of other rare bone diseases across Europe.

Political will and support at all governance levels (local, national, European/regional and international) are seen as crucial.