An ERN-BOND White Paper On Diagnosis

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



ERN-BOND

European Reference Network
for rare or low prevalence complex diseases
Network Bone Disorders (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.

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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

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- **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

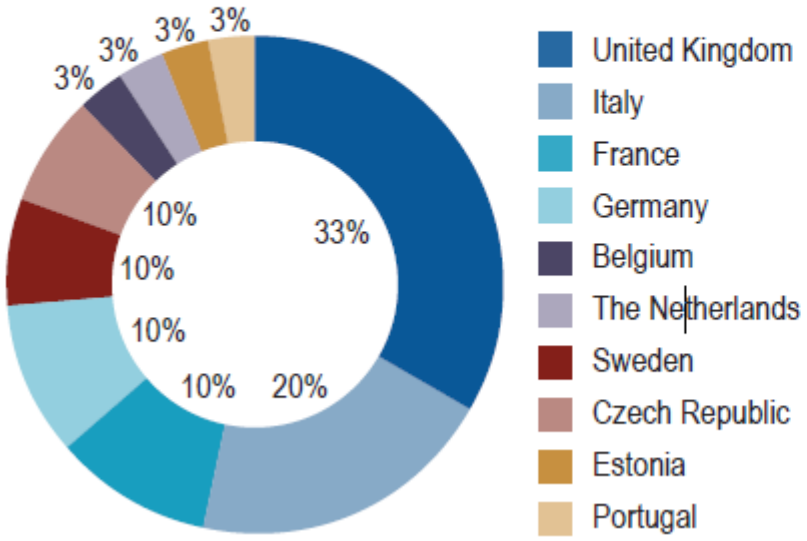
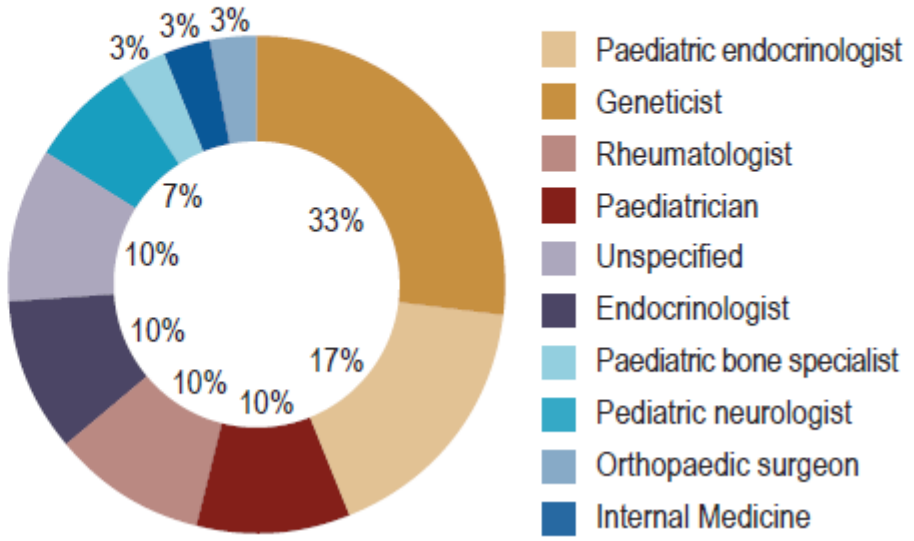


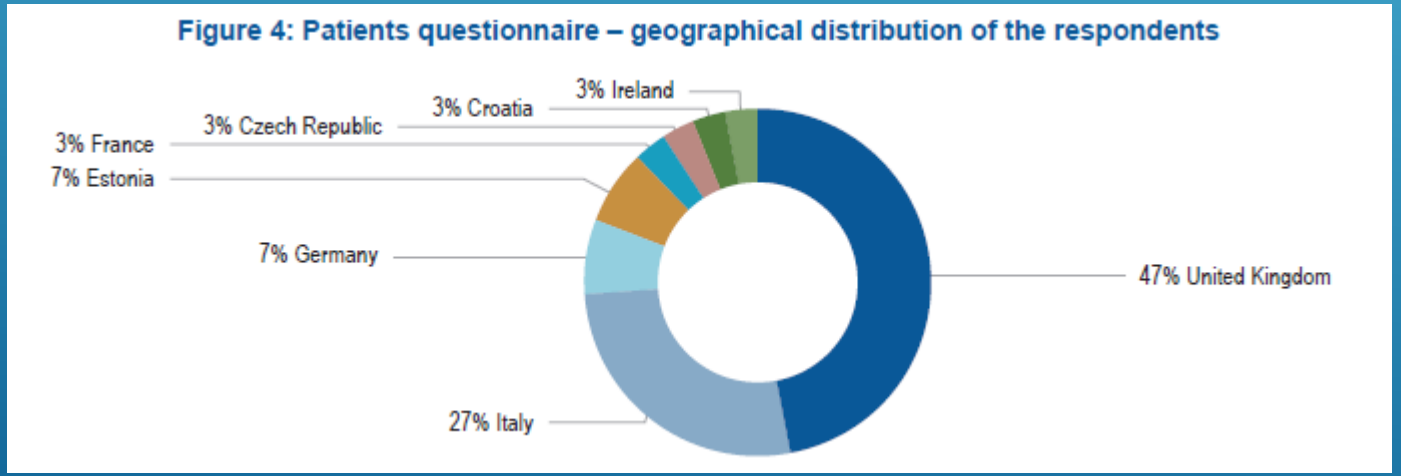
Figure 3: HCPs questionnaire – area of expertise of the respondents



THE SAMPLE: QUESTIONNAIRE FOR PATIENTS

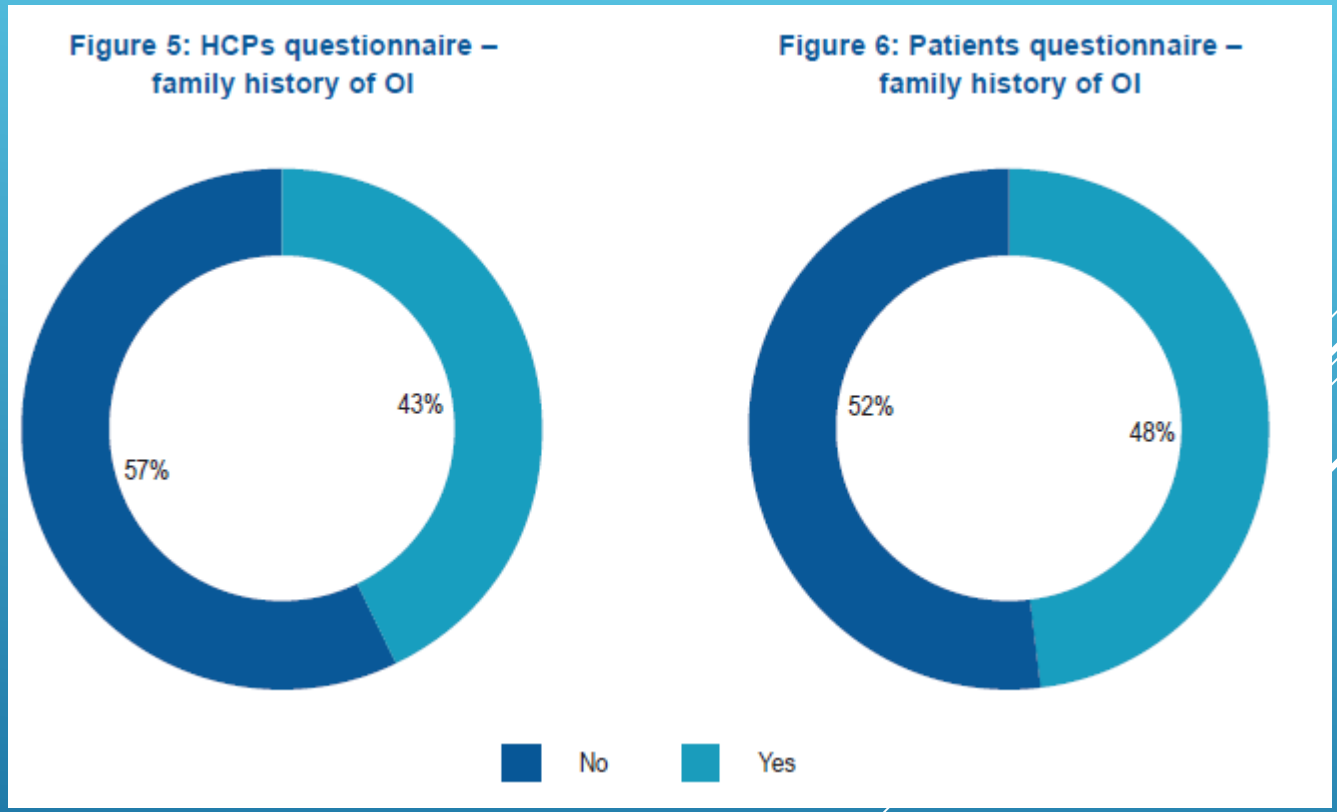
The age of the OI patients surveyed either directly or indirectly (via carers) varied greatly, ranging from new-borns to 71 years old.

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.



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.



DIAGNOSTIC PATHWAY

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

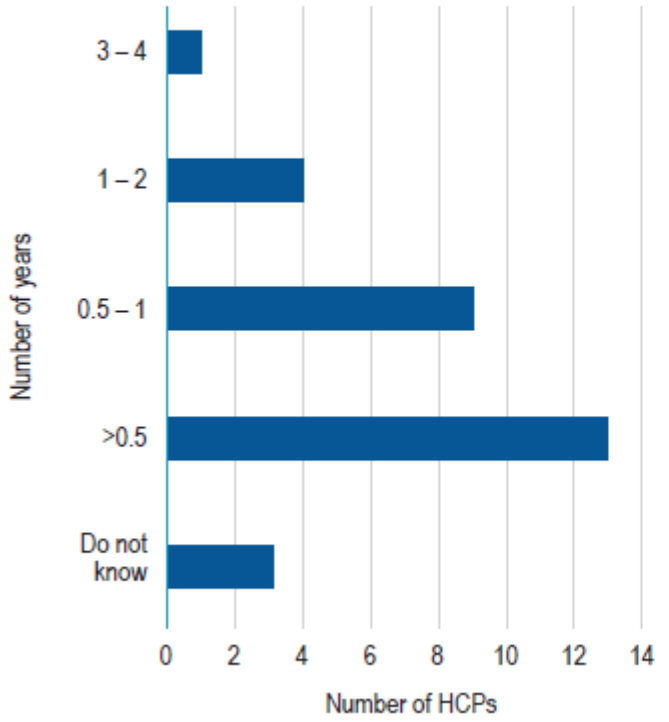
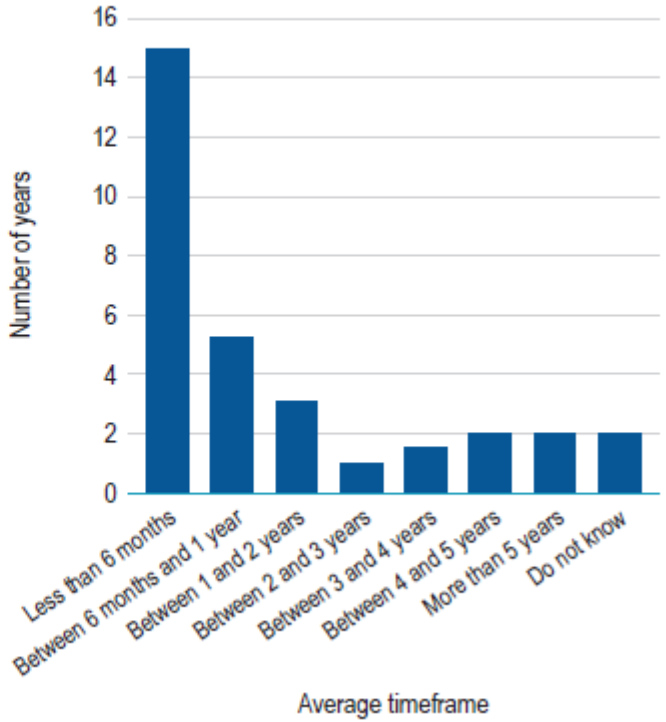


Figure 8: Patients questionnaire – average time-frame to receive diagnosis (in years)

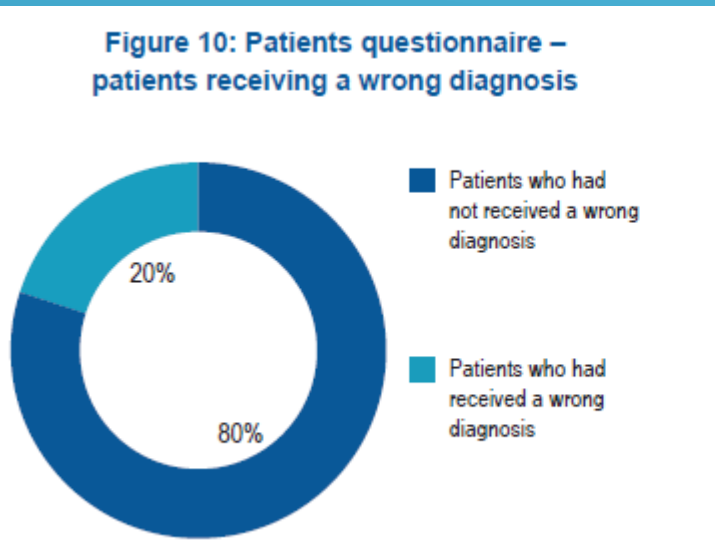


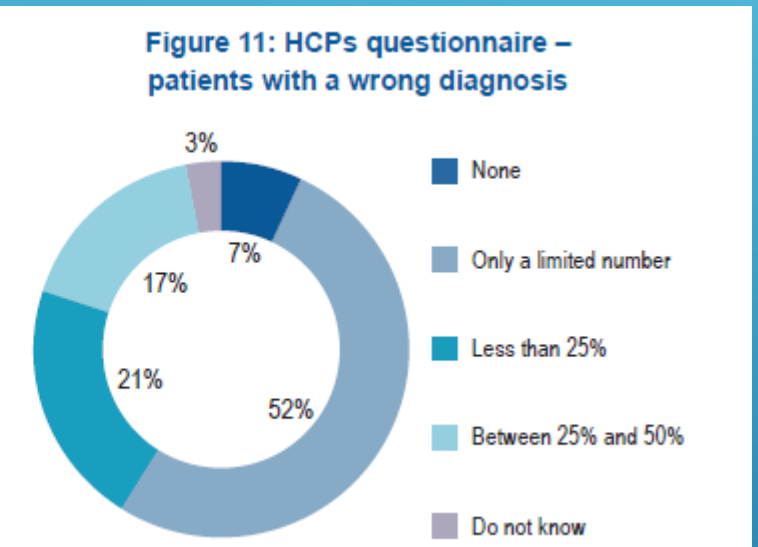
From the time of the disease's first symptoms emerging to obtaining an accurate diagnosis, the majority of both HCPs and patients indicated an average period of **less than 6 months to confirm the diagnosis.** This corresponds to **less than 5 visits between primary and specialised care.**

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





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 t o their Excellence Centre.

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

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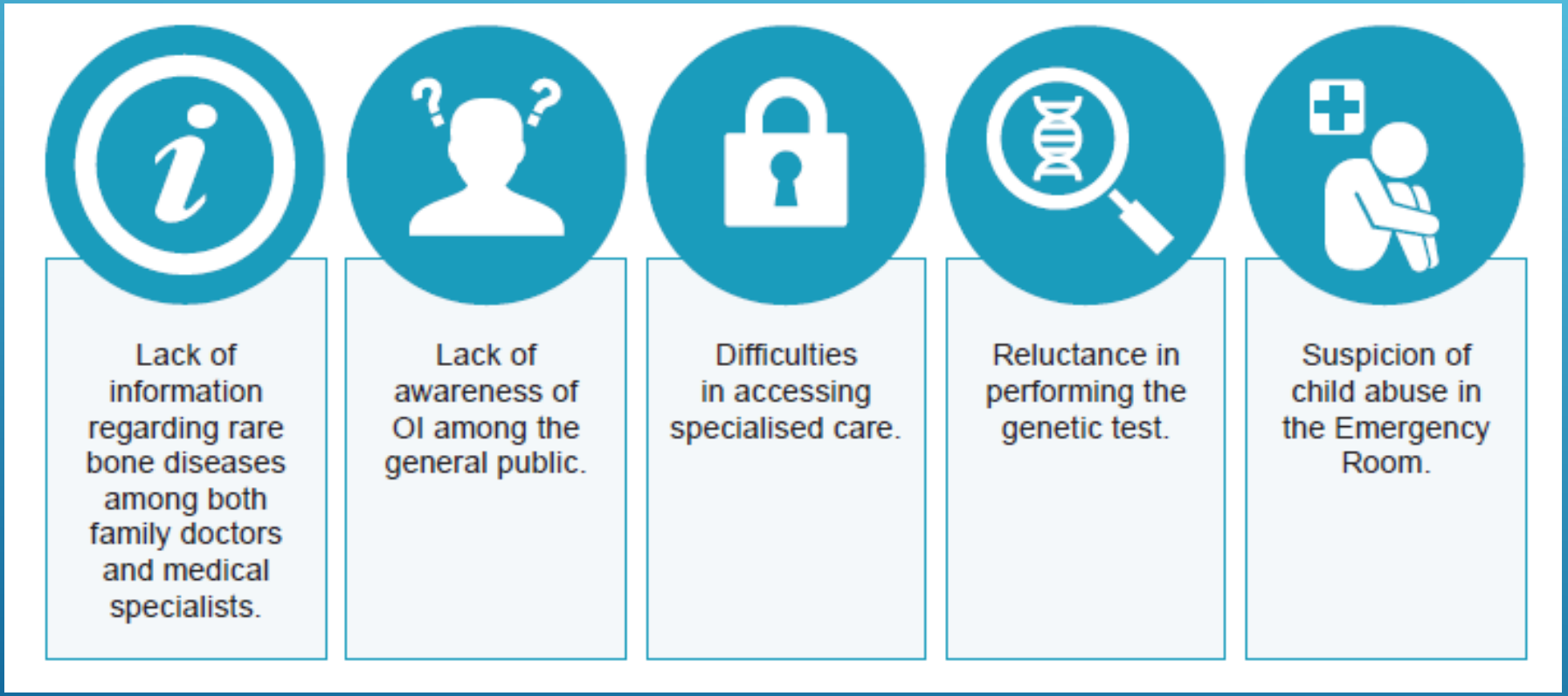
BARRIERS



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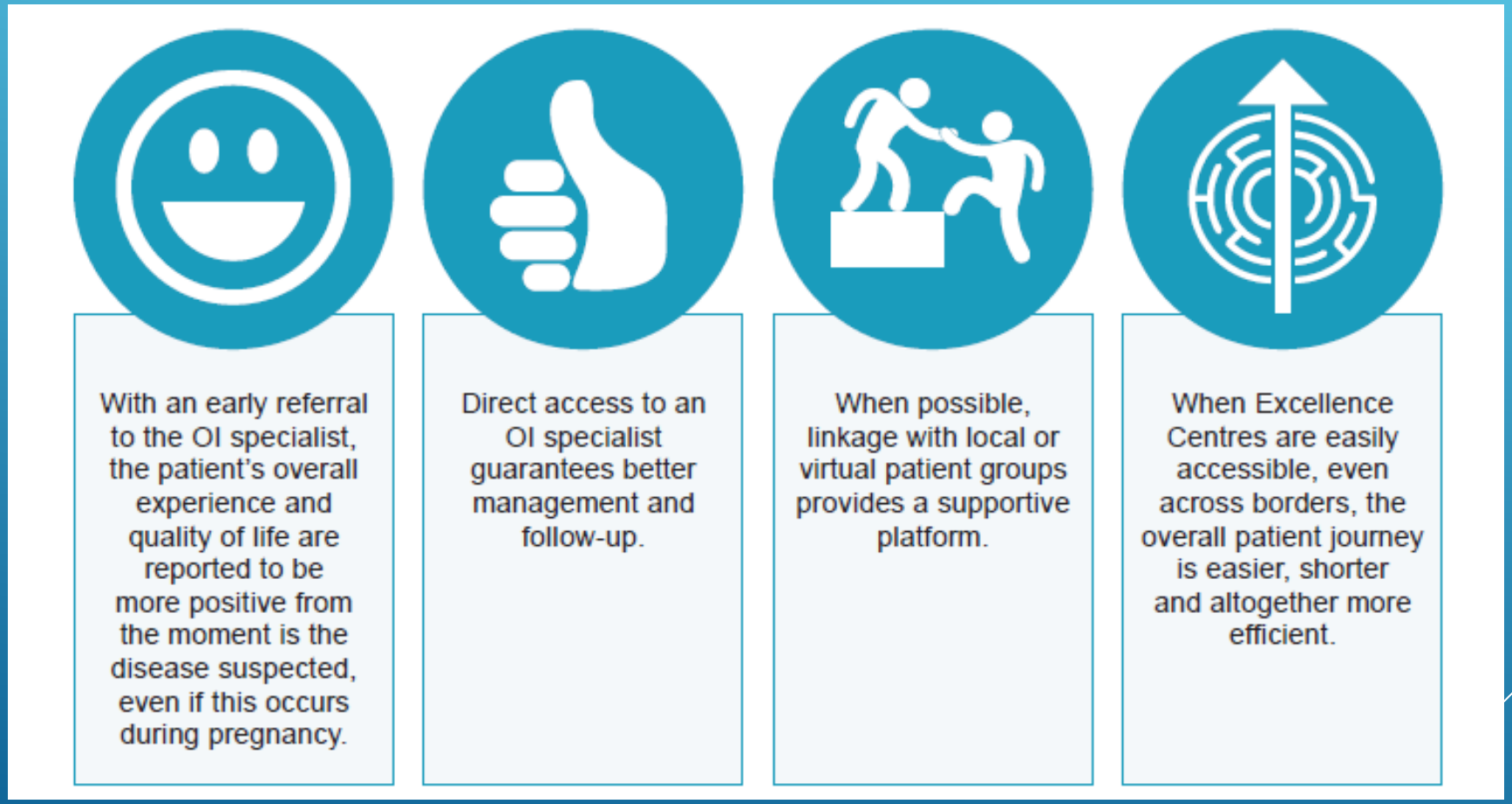
SUCCESS FACTORS



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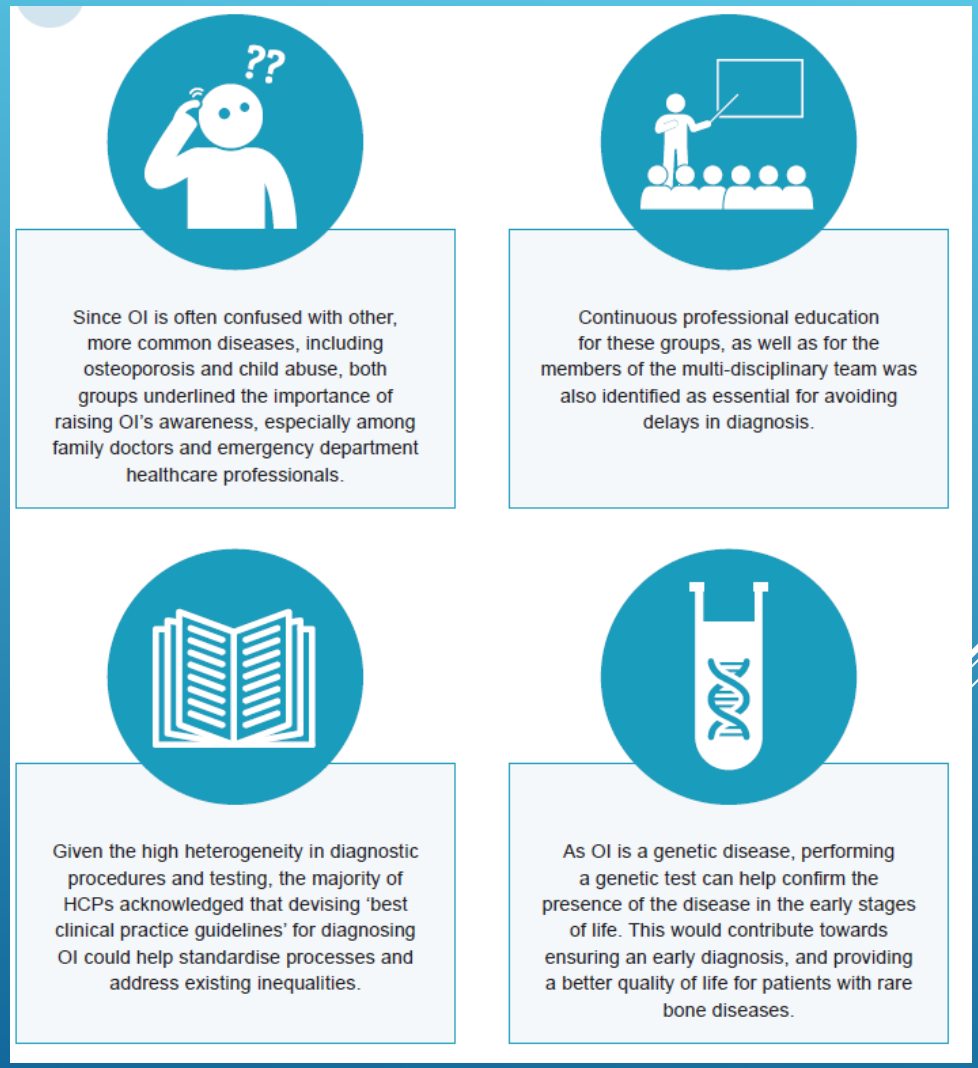
POTENTIAL SOLUTIONS

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.



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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.