

A European survey to identify new roads for care, training and research around rare metabolic bone diseases

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OBJECTIVES

As expertise in rare or low prevalence complex diseases or conditions is scarce and dispersed, the European Commission (EC) together with the Member States are working to establish European Reference Networks (ERNs) to link existing specialized healthcare providers (1). ERNs will facilitate access to diagnosis and treatment by centralizing knowledge, experience, medical research, training and resources in the area of rare or low prevalence complex diseases or conditions.

Rare metabolic bone diseases (RMBD) are at the crossways of Endocrinology, Nephrology, Orthopaedic surgery and Rheumatology. Most of RMBD lead to short stature, bone pain, tooth anomalies, leg deformities, bone fragility and disability of variable importance. The organization of care varies immensely amongst European countries. The announcement of the future organization of ERNs prompted us to conduct an international survey in order to identify and map the field of expertise, the organization of care and the current activities related to RMBD.

METHODS

A google questionnaire based on the operational criteria for Healthcare Providers (HCP) published by the European Commission (2) for the assessment of HCP applicants was sent to the existing networks; the information about the survey was conveyed through the ESPE, ECTS and ESE societies.

This survey consisted of 40 questions covering different themes such as the description of the local resources, the organization of care and the capacity to improve diagnosis and care through the development of guidelines and/or databases.

RESULTS

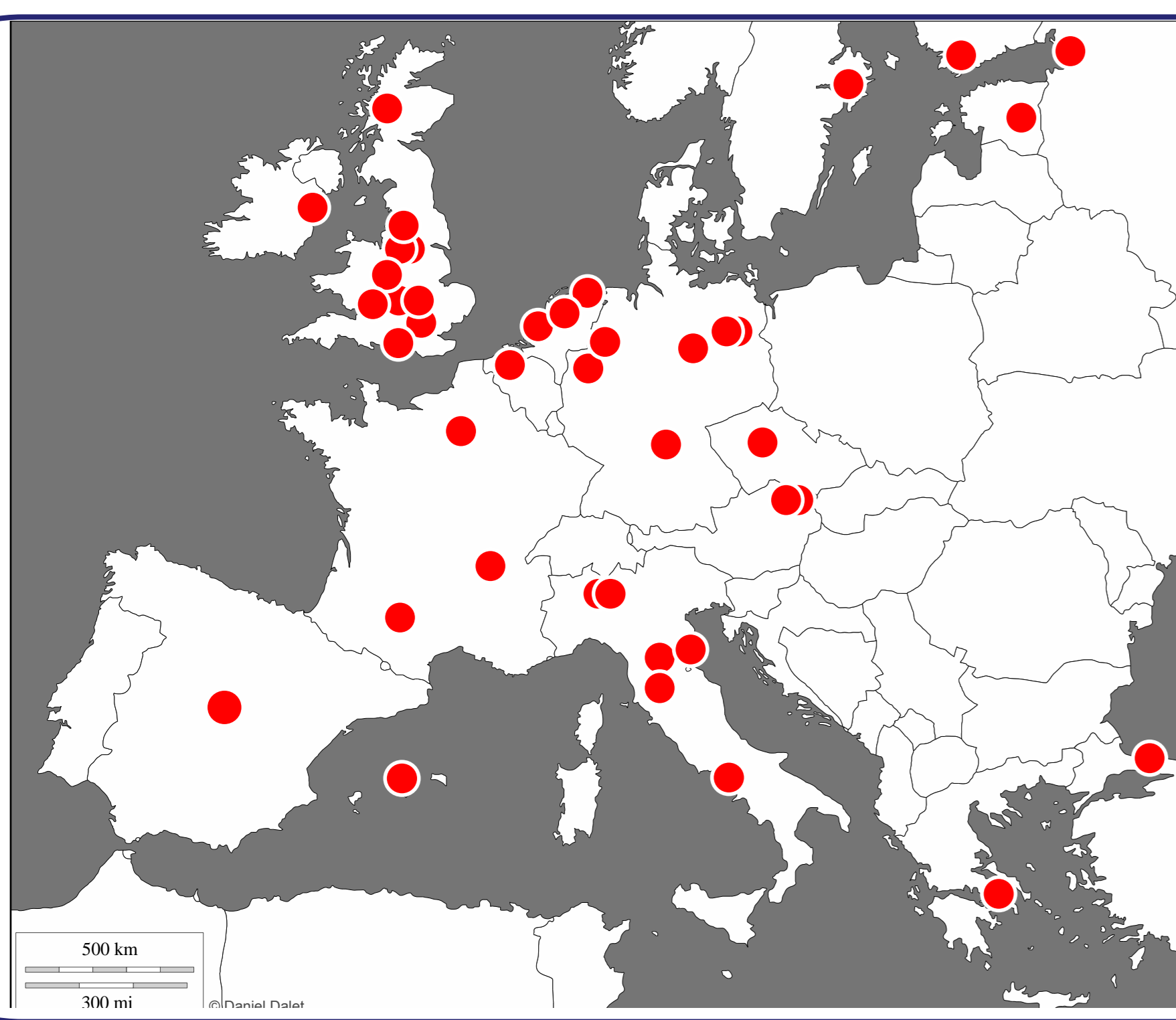


Fig 1: Geographical location of Healthcare Providers participating to the survey

A total of 42 centers across 19 European countries answered to the questionnaire. Each red dot represents one center of expertise.

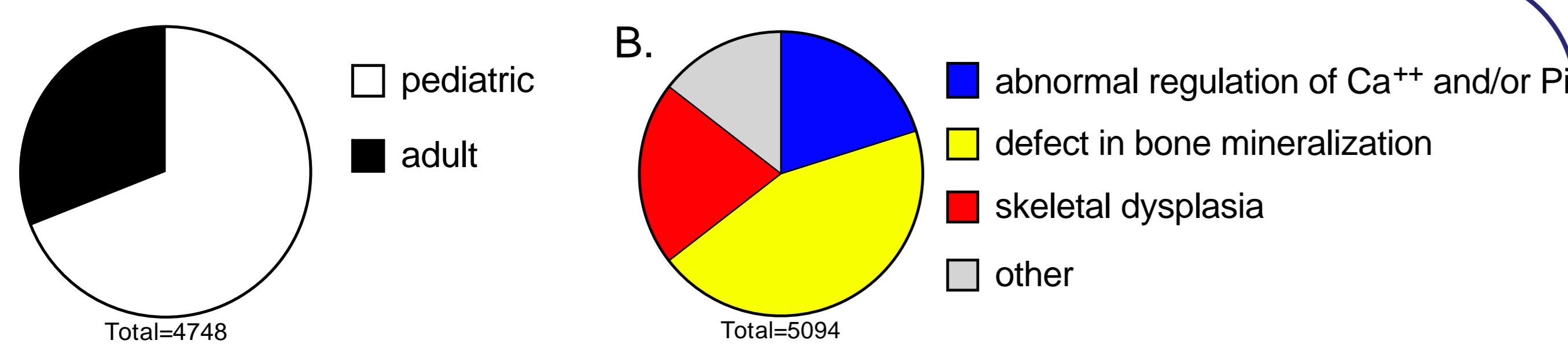
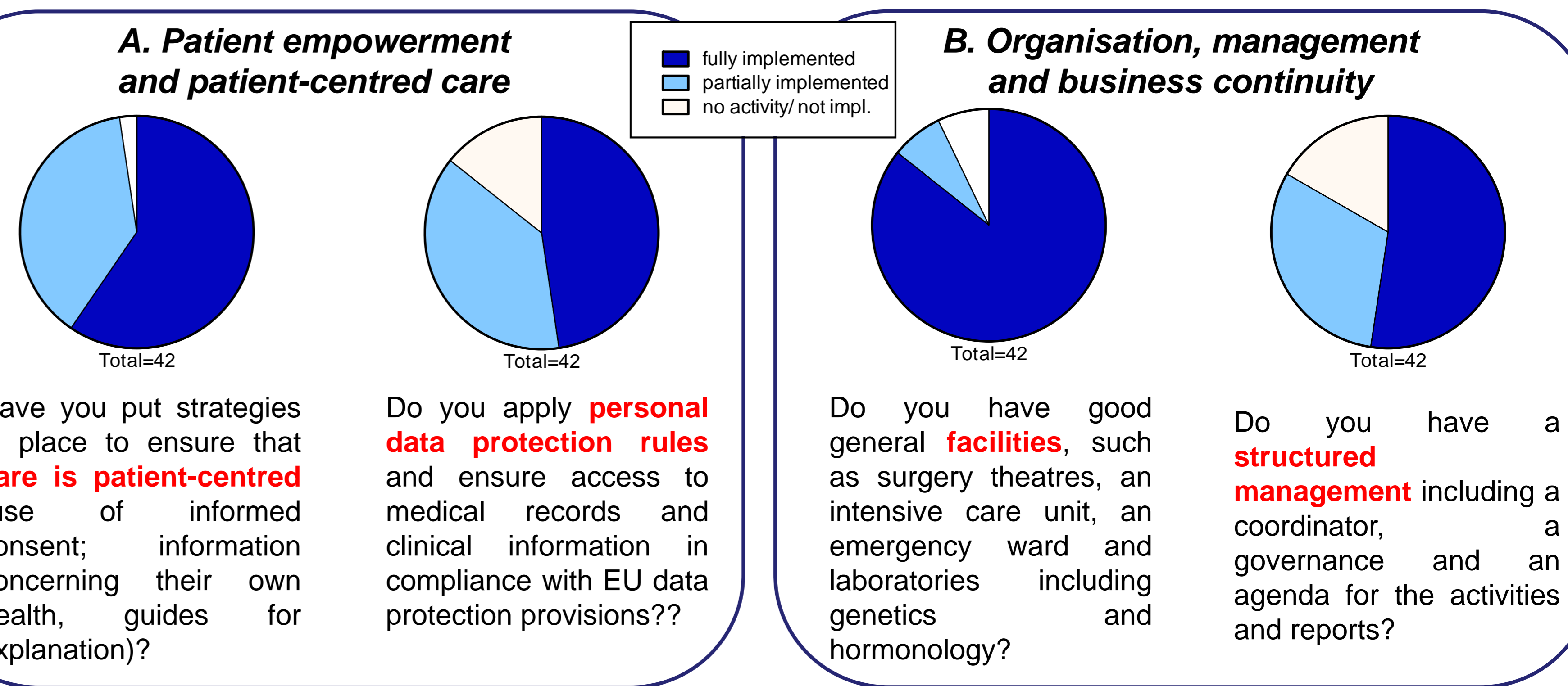


Fig 2: Rare Bone Diseases patients categories across all HCPs

A. Pie chart representing the total number of patients followed by the respondents to the survey (Pediatrics patients: n= 3275; adults patients: n= 1473).

B. Pie chart representing the different categories of patients treated by the respondents (abnormal regulation of Ca⁺⁺ and/or Pi; n= 1025, defect in bone mineralization: n=2262, skeletal dysplasia: n = 1069; others: n= 738).

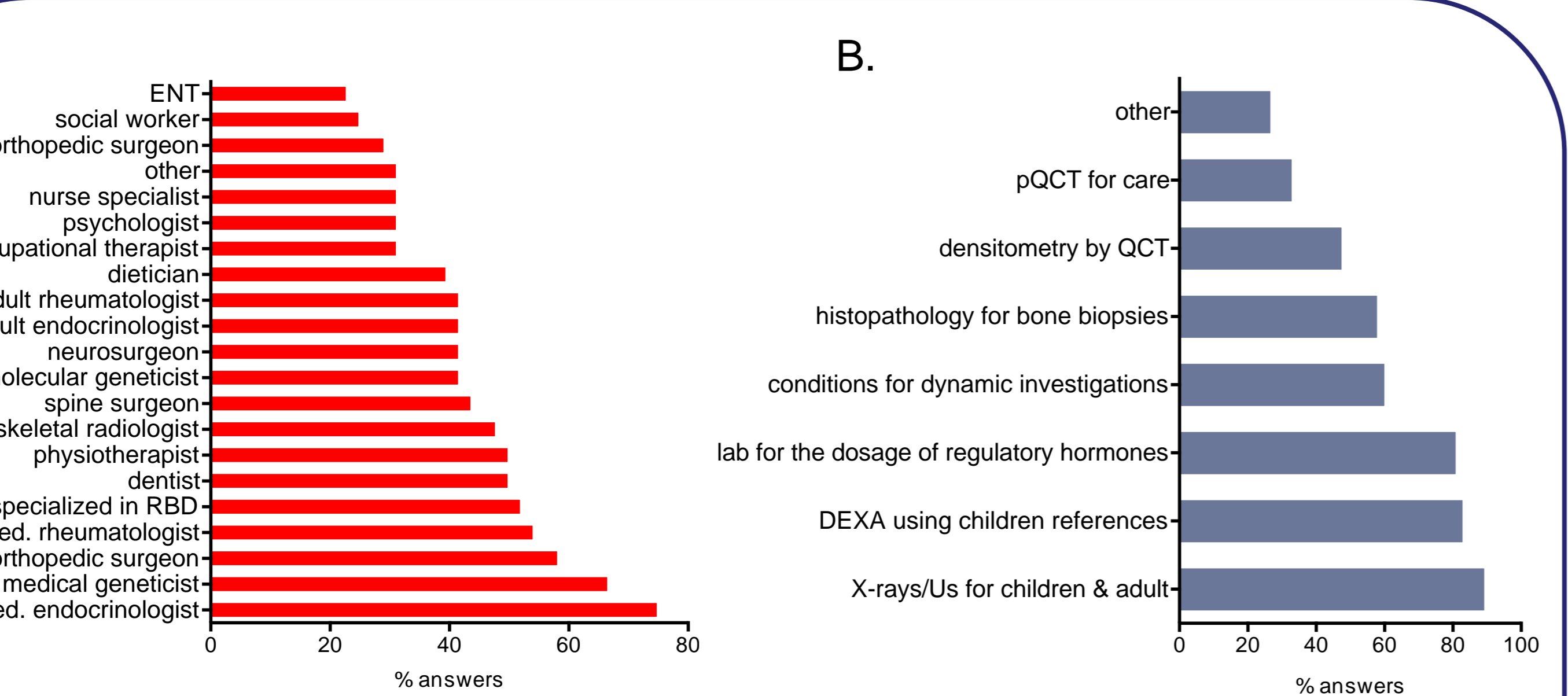
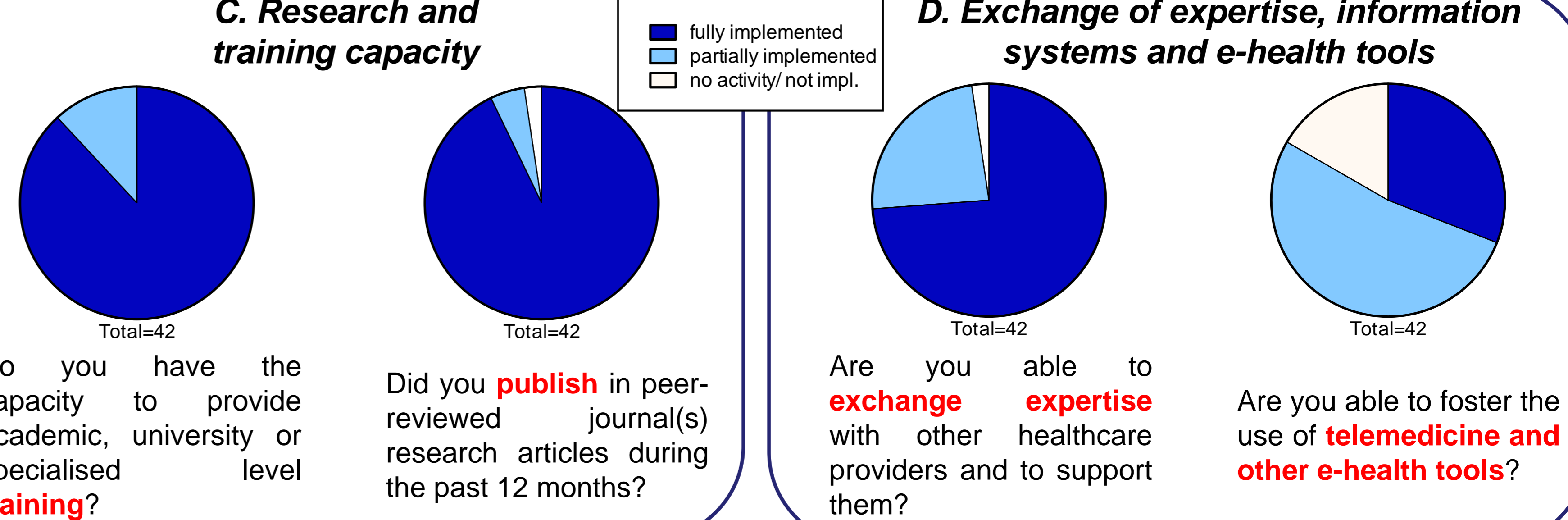


Fig 3: Characteristics of the local support

A. Medical resources dedicated to Rare Bone Diseases. Percentage of answers were calculated based on the total number of responses per question

B. Equipement or technical resources available for care (not research).

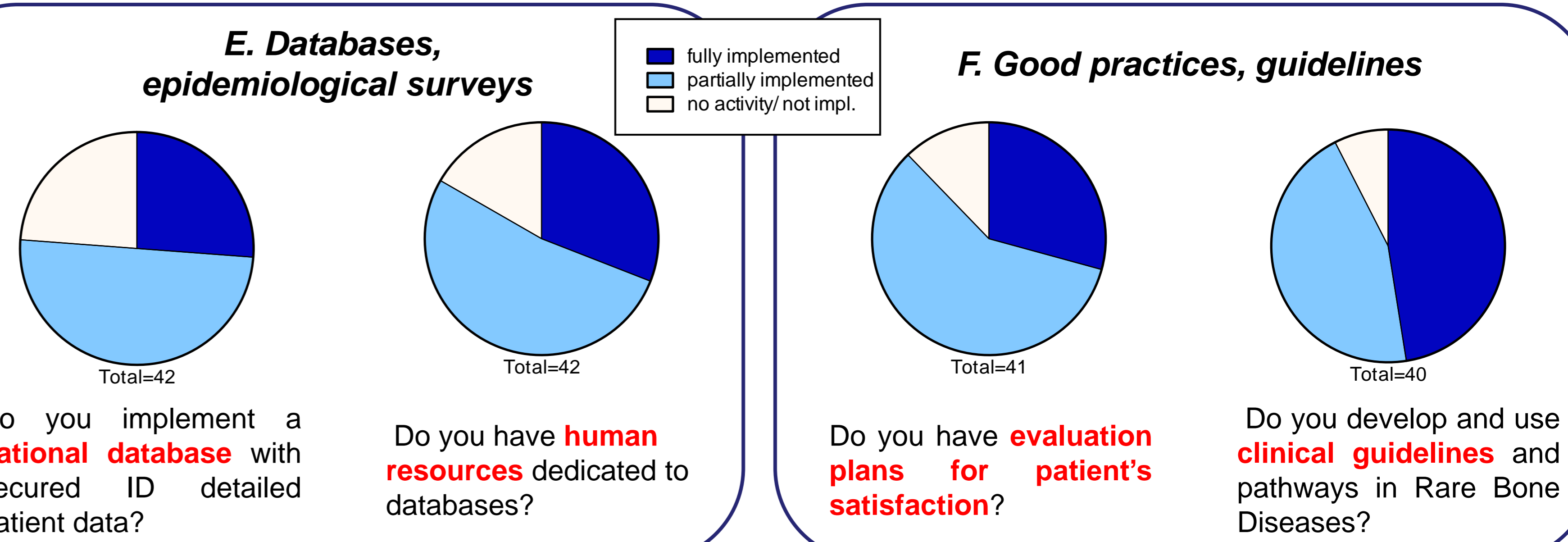


Fig 4 : Practices evaluation (panels A, B, C, D, E and F)

Healthcare professionals were asked to evaluate their practices with patients with RMBD. Pie charts represent the proportion of answers according to the level of implementation for each criterion (3 options were given: fully, partially or not implemented).

CONCLUSIONS

Our survey pointed the needs to 1- improve the visibility of expert centres towards patients and families, caregivers, and European health authorities; 2- guide the transition from child care to adult care; 3- harmonize care and enable clinical trials; 4- share the expertise and disseminate the knowledge of RMBD through e-tools and telemedicine. Our survey shows that European networking is a major instrument to improve the standards of care of patients affected with RMBD.

References

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2. http://ec.europa.eu/health/ern/docs/amt_operationalcriteria_healthcareprovider_en.pdf

Conflict of interest: the authors have no conflict of interest to report.