

Fernandez I¹, Ahmed SA², Argente J³, Bacchetta J⁴, Bianchi ML⁵, Bianchi, V⁶, Bishop N⁷, Boot AM⁸, Brandi ML⁹, Burren C¹⁰, Cheung M¹¹, Colao AM¹², Coucke P¹³, Davies J¹⁴, Diego de Sotto E¹⁵, Dougeraki A¹⁶, Gennari L¹⁷, Girschick H¹⁸, Grasemann C¹⁹, Fratzi-Zelman N²⁰, Haeusler G²¹, Hofmann C²², Hogler W²³, Javaid K²⁴, Kant S²⁵, Keen RW²⁶, Kopchak O²⁷, Kost M²⁷, Kostik M²⁷, Mäkitie O²⁸, McDonnell C²⁹, Mantovani G³⁰, Martos Moreno GA³, Mohnike K³¹, Nilsson O³², Oostdijk W²⁵, Padidela R³³, Salles JP³⁴, Schnabel D³⁵, Semler O³⁶, Sumnik Z³⁷, Tillmann V³⁸, Turan S³⁹, Shaw N²³, Usardi A¹, Walsh, J⁴⁰, Zillikens C⁴¹ and Linglart A^{1,42}.

¹. APHP, Plateforme Expertise Maladies Rares Paris Sud, Paris Sud, France, ².Glasgow, UK, ³.Madrid, Spain, ⁴.Lyon, France, ⁵.Milano, Italy, ⁶.San Marino, Italy, ⁷.Sheffield, UK, ⁸.Gröningen, The Netherlands, ⁹.Florence, Italy, ¹⁰.Bristol, UK, ¹¹.London, UK, ¹².Naples, Italy, ¹³.Ghent, Belgium, ¹⁴.Southampton, UK, ¹⁵. Balearic Islands, Spain, ¹⁶.Athens, Greece, ¹⁷.Siena, Italy, ¹⁸.Berlin, Germany, ¹⁹.Essen, Germany, ²⁰.Vienna, Austria, ²¹. Vienna, Austria, ²².Wuerzburg, Germany, ²³.Birmingham, UK, ²⁴.Oxford, UK, ²⁵.Leiden, The Netherlands, ²⁶.Stanmore, UK, ²⁷. St Petersburg, Russia, ²⁸. Helsinki, Finland, ²⁹.Dublin, Ireland, ³⁰.Milano, Italy, ³¹.Magdeburg, Germany, ³².Stockholm, Sweden, ³³.Manchester, UK, ³⁴.Toulouse, France, ³⁵.Berlin, Germany, ³⁶.Cologne, Germany, ³⁷.Prague, Czech Republic, ³⁸.Tartu, Estonia, ³⁹.Istanbul, Turkey, ⁴⁰.Sheffield, UK, ⁴¹.Rotterdam, The Netherlands, ⁴².Kremlin-Bicêtre, France.

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

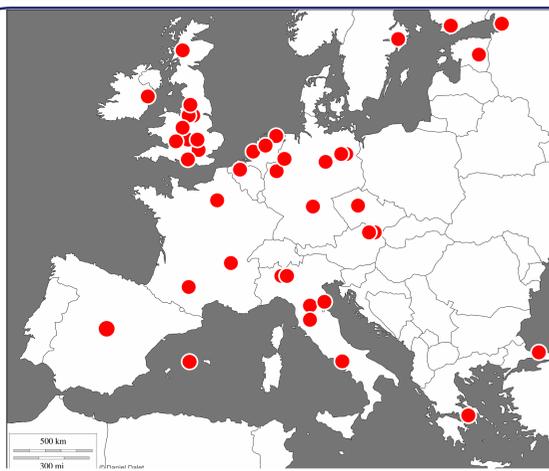
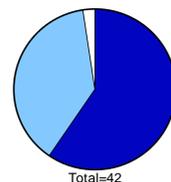


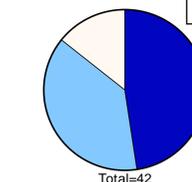
Fig 1: Geographical location of Healthcare Providers participating to the survey

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

A. Patient empowerment and patient-centred care

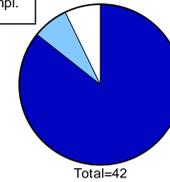


Have you put strategies in place to ensure that **care is patient-centred** (use of informed consent; information concerning their own health, guides for explanation)?

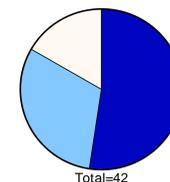


Do you apply **personal data protection rules** and ensure access to medical records and clinical information in compliance with EU data protection provisions??

B. Organisation, management and business continuity



Do you have good general **facilities**, such as surgery theatres, an intensive care unit, an emergency ward and laboratories including genetics and hormoneology?



Do you have a **structured management** including a coordinator, a governance and an agenda for the activities and reports?

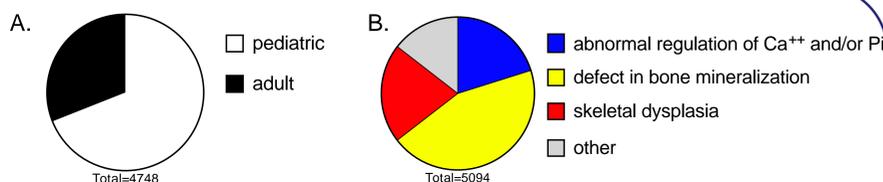
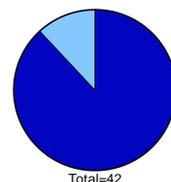


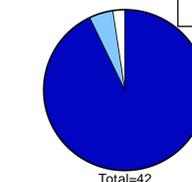
Fig 2: Rare Bone Diseases patients categories across all HCPs

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).

C. Research and training capacity

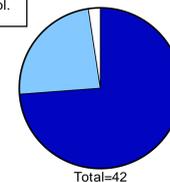


Do you have the capacity to provide academic, university or specialised **training**?

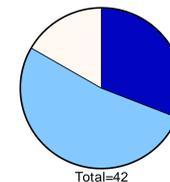


Did you **publish** in peer-reviewed journal(s) research articles during the past 12 months?

D. Exchange of expertise, information systems and e-health tools



Are you able to **exchange expertise** with other healthcare providers and to support them?



Are you able to foster the use of **telemedicine and other e-health tools**?

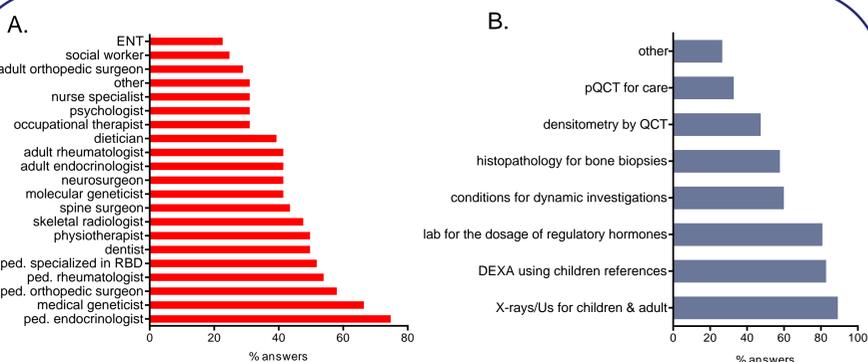
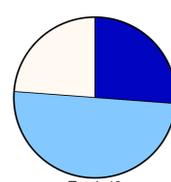


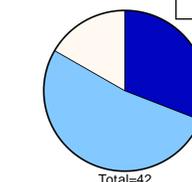
Fig 3: Characteristics of the local support

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).

E. Databases, epidemiological surveys

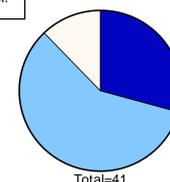


Do you implement a **national database** with secured ID detailed patient data?

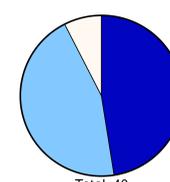


Do you have **human resources** dedicated to databases?

F. Good practices, guidelines



Do you have **evaluation plans** for patient's satisfaction?



Do you develop and use **clinical guidelines** and pathways in Rare Bone Diseases?

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.