**European Reference Networks (ERNs) Accelerating and Improving Diagnosis for Rare Disease Patients**

On the occasion of Rare Diseases Day, on 28 February 2018, the European Reference Network for Rare Bone Disorders (ERN-BOND) coordinated by Luca Sangiorgi (IT), with the kind support of EURORDIS, organized a European Parliamentary Meeting entitled: **European Reference Networks – Accelerating and Improving Diagnosis for Rare Diseases Patients**. The meeting was kindly hosted by MEP Elena Gentile (S&D, IT).

The event was aimed to present the findings of the ERN-BOND White Paper on Diagnosis and discuss solutions on how to best reduce the average time to accurate diagnosis for rare bone diseases and, more generally, rare diseases.

Dr. Vytenis Andriukaitis, European Commissioner for Health and Food Safety, was one of the invited speaker to participate in the meeting to celebrate one year of success of the 24 ERNs that are now operating across Europe. In his talk he expressed grateful for the continuous work that ERNs are doing for patients, alongside healthcare professionals, researchers, policy makers and hospital managers – working together to improve the quality of life of millions of people in Europe suffering from a rare disease.

He also highlighted three immediate challenges for the future of the ERN:

1. to ensure the integration of ERNs into national or regional healthcare systems.
2. to ensure full support to the ERNs
3. the selection and integration of new members in the 24 current ERNs.

To read the full speech of Dr. Vytenis Andriukaitis click [here](https://www.dropbox.com/s/23yfgbcmhdbln1d/Speech%20Commissioner%20%282%29.pdf?dl=0)

Importantly during the meeting the results of the ERN-BOND white paper on diagnosis were released. The white paper provides an overview of the current situation relating to the diagnosing of osteogenesis imperfecta (OI) in the 10 Member States represented within the ERN-BOND.

OI, also known as brittle bone disease, is a genetic disorder that affects mainly bones caused by a qualitative or quantitative defect in type I collagen, which is sometimes associated with extreme bone fragility and an increased risk of fractures. Results show the need for better awareness of rare diseases.

The objective of the White Paper therefore consists in identifying the key challenges and potential solutions to further reduce inequalities in OI diagnosis between countries and delays in detection and improve the patient experience. The white paper provides an overview of the state of play of OI diagnosis in the centres that participated in the survey. Even though the results do not reflect the situation in the whole

The ERN-BOND White Paper says, “The challenges in diagnosing OI directly impacts patients as delays can lead to inappropriate or delayed management or even unnecessary interventions.”

Due to the relatively lack of awareness of the disease not just among patients and families but also among some health practitioners themselves (doctors, nurses or emergency personnel), symptoms are sometimes not recognized and confused with those of child abuse. This dramatically impacts patients as delays can lead to inappropriate or delayed management, or even unnecessary interventions.

The paper identifies the following 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.

1. Education and training Awareness-raising activities on rare bone diseases
2. Creation of national clinical networks connected to the Excellence Centres
3. Development of European guidelines for OI diagnosis
4. Empowering patients and their carers through development of local support groups

In the second part of the meeting

* Luca Sangiorgi, ERN-BOND Coordinator, Istituto Ortopedico Rizzoli, Italy
* Maurizio Scarpa, Chair, ERN Coordinators Group
* Yann Le Cam, Chief Executive Officer, EURORDIS- Rare Diseases Europe
* Martin Seychell, Deputy Director General of the Directorate-General for Health and Food Safety

Took part in the Panel discussion entitled **How to address the challenges of early diagnosis of rare diseases** **moderated by Josep Figueras, Director of the European Observatory on Health Systems and Policies.**

From the discussion it appeared that political intention and support at all governance levels (local, national, European/regional and international) is seen as crucial for prioritising the key issues of early diagnosis of Rare Diseases.