ERN on bone disorders (ERN BOND)

Rare bone diseases encompass disorders of bone formation, modelling, remodelling and removal, and defects of the regulatory pathways of these processes. They result in short stature, bone deformity, teeth anomalies, pain, fractures and disability, and can adversely influence neuromuscular function and haemopoiesis.

ERN BOND brings together all rare bone diseases — congenital, chronic and of genetic origin — that affect cartilage, bones and dentin. The network is focusing initially on osteogenesis imperfecta (OI), X-linked hypophosphataemic rickets (XLH) and achondroplasia (ACH) as exemplars, based on disease prevalence, diagnostic and management difficulty and novel emergent therapy, before moving on to rarer diseases when systematic approaches are established.

Working with patients, BOND will develop patient-reported outcome and experience measures. The network will develop guidelines, leading to the development and dissemination of best practice. As new therapeutics are developed, the network will work to ensure rapid access to studies for affected patients.

BOND will enable skill development through eHealth and telemedicine platforms, alongside working visits, training courses and dissemination activities. The network aims to reduce time to diagnosis with fewer inappropriate tests, more accurate diagnosis and new viable treatments to be available within 2 to 3 years.

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NETWORK COORDINATOR

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