Question for written answer E-001106/2018
to the Commission
Rule 130
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Subject: XLH within ERNs

X-Linked Hypophosphatemia (XLH) is a rare, chronic and progressive musculoskeletal disorder, with an estimated incidence rate of 1 per 20 000 to 25 000 live births. Children born with the disorder may suffer from lower-extremity deformities, while all patients may have a seriously impaired quality of life due to skeletal disease and lifelong chronic pain.

Conventional treatment for XLH, consisting of multiple daily doses of vitamin D/phosphate supplements, is inadequate and ineffective. Recently, significant improvements have been made in delivering effective treatment, yet a lack of knowledge about the disease among healthcare professionals persists.

The European Reference Networks (ERNs) are an important tool for the EU in improving the situation for rare disease patients. XLH has been singled out as one of the preliminary diseases to be prioritised by the ERN on Rare Bone Disorders (ERN-BOND), in collaboration with the ERN on Rare Endocrine Conditions (Endo-ERN).

1. What steps can the Commission take to make sure that additional awareness is raised regarding XLH, especially among clinicians?

2. How can the Commission ensure that ERNs, especially where interconnected, are optimised to aid patient identification efforts so as to reach patients as early as possible in the course of their disease?