Real-world Clinical Profiles of Children With Hypophosphatasia From the Global HPP Registry

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INTRODUCTION

- Hypophosphatasia (HPP) is a rare, inherited, metabolic disease caused by deficiency of tissue-nonspecific alkaline phosphatase (TNAP) that can affect almost every system of the body.
- Classically, HPP is considered a disease primarily of the skeletal system3,4; however, severe disease manifestations, including disease signs and symptoms, age at first treatment1,2,5,6

METHODS

- The Global HPP Registry, an observational, prospective, multinational study (NCT01908470; EUPAS431092), was established in 2013 to enable healthcare practitioners to collect data on patient demographics and medical history related to HPP including disease signs and symptoms, age at first manifestation, and ERT treatment status.

RESULTS

- Patients <10 years of age at baseline from the Global HPP Registry were included in this study if they had:
  - Low serum alkaline phosphatase activity (ALP) activity under the lower limit of the age- and sex-adjusted reference range with no other cause for hyperphosphatemia
  - Valid enrollment date, date of enrolment or date of birth, known sex, mode of family, known ERT treatment status (treated or untreated), and treatment start date if ever treated
  - History of one or more prespecified signs or symptoms of HPP at baseline

- Other criteria:
  - History of specific signs and symptoms of HPP within each body system at baseline

CONCLUSIONS

- The Global HPP Registry is observational. Baseline data and retrospective medical records were collected and reviewed to reflect variations in the reported severity of the disease from different countries and in the standard of care for the disease.
- Most patients had multiple organ system involvement involving both severe disease manifestations and chronic signs and symptoms.

Most common signs and symptoms of HPP in children ≥2 baseline signs and symptoms, excluding those presenting with only dental manifestations (n=165)

**REFERENCES**

- Genetics of Bone Biology and Stock options in the company.
- Conclusions of HPP Registries:

- - Regional differences in clinical presentation were noted.
- - Japan had the largest reported population of patients diagnosed at <6 months, largest percentage of patients with skeletal and respiratory manifestations, and a higher percentage of patients in the 0–12 month age group.
- - The United States/Canada had the largest percentage of patients with skeletal and respiratory manifestations.
- - Europe had the largest percentage of patients with renal and respiratory manifestations.
- - A comparison of HPP signs and symptoms in children with varying baseline characteristics revealed that most treated the importance of understanding differences in clinical presentation of HPP, particularly differences based on geographical region.

- Additionally, non-stated manifestations, such as hearing loss, growth failure, motor delay, fatigue, and failure to thrive, commonly occur and should be considered in the diagnosis and the decision to treat HPP patients with ERT.

- These findings may be used as a primary tool for assessment of symptoms and actual activity versus disease progression in HPP.

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**DISCLOSURES**


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