INTRODUCTION

Hypophosphatasia (HPP) is a rare, inherited, systemic, metabolic disease characterized by early-onset pathology due to impaired mineralization or osteogenesis.

Objective

The objective of this study was to describe the clinical, biochemical, and radiographic features of patients with HPP using a large electronic health record (EHR) database.

Methods

Database

We identified potential patients with HPP using an EHR database of an academic medical center. The study period was August 1, 1991, to December 31, 2014.

Study Definitions (Figure 1)

- Index date: the date of diagnosis of disorder of phosphorus metabolism and/or the second low ALP activity finding (majority of ALP values low)
- Date of diagnosis of disorder of phosphorus metabolism or the second finding of a low ALP activity level
- Period of diagnosis: the period from the index date to the date of diagnosis, or the second finding of a low ALP activity level
- Time from first activity date to subsequent fractures
- Time from index date to subsequent fractures
- Post-index period (i.e., follow-up) varied and had no minimum duration but did not extend beyond the enrollment period

Results

A total of 301 patients were included in the cohort, with 295 patients (98.3%) meeting the clinical, biochemical, and radiographic criteria for HPP.

Baseline characteristics of the patients are shown in Table 1. The mean age at diagnosis was 8.0 years (SD 7.3), and 53.2% of patients were male.

Clinical, biochemical, and radiographic evidence of HPP noted by ≥1 of the following:

- Elevated urine phosphoethanolamine
- Bone pain
- Fractures
- Radiographic evidence of hypomineralization, osteopenia, or rickets-like features
- Persistently low age-/sex-adjusted ALP activity noted by CPT 84705 (≥2 measurements, low/never normal activity levels, and no bisphosphonate use)
- Family history of HPP
- Certain HPP complications
- Craniosynostosis
- Respiratory failure
- Premature birth
- Other anomalies

CONCLUSIONS

This study describes the clinical, biochemical, and radiographic features of a large cohort of patients with HPP identified within an electronic health records database of an academic medical center.

REFERENCES


DISCLOSURES

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