A Health System-Based Approach: Using EMR Filters to Identify Suspected Occult Hypophosphatasia

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INTRODUCTION: Occult hypophosphatasia (HPP) is a rare metabolic bone disorder caused by mutations in the ALPL gene; the prevalence of severe forms of the disease has been estimated to be 1/100,000. It is characterized by a wide variety of clinical symptoms including a persistently low tissue-non-specific alkaline phosphate (ALP). ALP is important for mineralization of the bones and teeth. Therefore, HPP can have serious consequences if left undiagnosed. Identifying patients with HPP can be challenging because a low ALP is uncommonly encountered in clinical practice and HPP has a nonspecific clinical presentation in milder forms. Electronic medical record (EMR) systems have become an integral part of patient care, and there is growing interest in their potential to improve disease surveillance and early diagnosis. The purpose of this study is to investigate the use of EMR filters in the identification of potential cases of occult HPP in a large, rural health system.

METHODS: After IRB approval, our Information Technology department utilized filters to identify adult patients (>18) who had visited the health system from 2014 to 2018 and had two consecutively low ALP levels (serum alkaline phosphatase ≤30 IU/L). Patient charts that met the search criteria were then reviewed by the research team to confirm the potential diagnosis and collect data on their clinical characteristics. Exclusion criteria for patients included the presence of diseases and treatments known to decrease ALP levels, such as active cancer, multiple myeloma, overt hypothyroidism, malnutrition, pernicious or profound anemia, multiorgan/hepatic failure, bisphosphonate therapy, chronic glucocorticoids, and ongoing chemotherapy.

RESULTS: Of the 733,860 unique patients seen between 2014 and 2018, 273 patients were identified with two consecutively low ALP levels. Among these patients, 28 were identified after chart review as potentially having occult HPP. Common symptoms reported included musculoskeletal pain, recurrent fractures, myalgia, chondrocalcinosis, and dental problems, such as periodontal disease.

DISCUSSION: In conclusion, the utilization of EMR filters to identify potential cases of occult HPP has shown to be an effective approach; by leveraging the power of EMRs, healthcare providers can quickly identify patients at risk for HPP and initiate appropriate interventions. Going forward, the next step would be to contact the identified patients for further evaluation and management. By taking a proactive automated approach to screening and diagnosis, outcomes can potentially be improved for patients with HPP.

SIGNIFICANCE/CLINICAL RELEVANCE: This study highlights the successful use of EMR filters to identify potential cases of occult HPP within a large health system, aiding in the prompt recognition and management of this rare metabolic bone disorder. Leveraging EMRs for proactive screening could improve outcomes by enabling timely interventions for patients with HPP symptoms and low ALP levels.

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IMAGES AND TABLES

Figure 1: Methodology

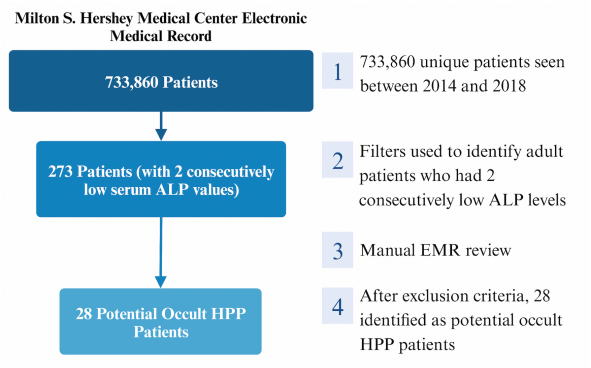

Table 1: Exclusion criteria for manual EMR review

Diseases and treatments known to decrease ALP levels	
Active cancer	Multiorgan failure
Multiple myeloma	Hepatic failure
Overt hypothyroidism	Bisphosphonate therapy
Malnutrition	Chronic glucocorticoids
Pernicious/profound anemia	Ongoing chemotherapy

Table 2: Clinical manifestations of adult occult hypophosphatasia

Clinical manifestations	
Neurological	Seizures (Vit. B6 responsive)
Skeletal	Defective mineralization, rickets, osteomalacia, bone deformity, multiple fractures, delayed bone healing
Respiratory	Severe respiratory insufficiency, respiratory complications
Muscular	Chronic muscle pain, reduced muscular strength and performance
Articular	Chondrocalcinosis, ectopic calcification of ligaments, pseudogout
Renal	Hyperphosphatemia, hypercalciuria, nephrocalcinosis
Dental	Premature loss of decidual and permanent teeth, periodontal disease