SHORT COMMUNICATION

IDENTIFYING INDIVIDUALS AT RISK FOR HYPOPHOSPHATASIA USING AN ELECTRONIC MEDICAL RECORD (EMR)

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Purpose
Utilizing preventive medicine management tools in an Electronic Medical Record (EMR) serving 3.3 million Kaiser Permanente beneficiaries in Southern California, we sought to identify individuals with possible hypophosphatasia (HPP).
Background
Kaiser Permanente HealthConnect (HC) is an integrated EMR using the "Epic" system, allowing secure access to provider notes, diagnoses, laboratory, radiology, and pharmacy records serving approximately 10 million beneficiaries in the United States. Preventive Medicine applications allow searches by ICD-9 diagnosis code and abnormal laboratory parameters. HPP in its homozygous form is a severe metabolic bone disease with prevalence estimates ranging from 1/100,000 to 1/300,000 in populations of European origin. In its heterozygous state, HPP may be a more subtle disorder presenting with bone pain and stress fractures. It has long been postulated that mild forms of HPP are more prevalent in an adult population than currently recognized. Based on molecular genotype/phenotype correlations, an estimated prevalence fro moderate HPP of ~ 1/6370 has been proposed in Europe.
Methods
A pilot project was felt by the Institutional Review Board to fall within the confines of preventive care. The EMR of ~ 500,000 San Diego county Kaiser Permanente beneficiaries were queried for serum alkaline phosphatase (ALP) values less than the age-related normal values in the laboratory system (e.g. for adults, ≤ 20 IU/L with normal having been defined 20-125 IU/L). These records were cross-referenced to a list of ICD-9 diagnostic codes associated with manifestations of HPP. Radiographic reports and in many cases, the actual radiographs themselves could be reviewed for manifestations of HPP. The likelihood of mild to moderate HPP was assessed from the available clinical, biochemical and radiographic evidence in the EMR. Identified at risk individual's primary care providers were contacted to suggest confirmatory studies.
Results
94 individuals were identified in whom serum ALP was persistently low with at least one value recorded ≤ the age-related norm (e.g. < 20 IU/L). 29 individuals had recorded ICD-9 diagnoses which made HPP possible, including 1 pediatric patient and 3 adult patients who already carried the diagnosis. Of these 25 remaining individuals, 9 have thus far had biochemical confirmation of mild to moderate HPP, none have thus far been characterized from a molecular standpoint.
Conclusions
In spite of clear issues with bias of ascertainment, the EMR can be readily used to identify individuals at risk for HPP not previously identified by their clinicians. Further clinical and biochemical investigation of the likely affected individuals is being pursued. IRB guidance in expanding to the larger HC served population is being sought.