

How the precision of real-world lab data transforms rare disease patient identification

Comparing claims data vs lab data in identifying patients with hypophosphatasia

Hypophosphatasia (HPP) traps patients in a prolonged diagnostic odyssey because its vague symptoms are frequently misdiagnosed as osteoporosis and/or fibromyalgia. Relying solely on claims data fails to find these hidden patients, which significantly delays proper care and exposes them to contraindicated treatments. **However, integrating diagnostic lab data cuts through misleading claims history to reveal the true clinical markers of the disease.**

Analysis

Quest data scientists analyzed both claims and lab data to demonstrate how the precision of clinical lab results surpasses diagnosis codes in identifying patients with hypophosphatasia who had been misdiagnosed with other musculoskeletal diseases.

Specifications

Patients analyzed:

- Patients with a claims diagnosis code for osteoporosis with and without current pathological fracture (ICD-10: M80.0 and M81.0), fibromyalgia (ICD-10: M79.7), or other specified disorders of bone density and structure (ICD-10: M85.8X)
- Patients with a claims diagnosis code for hypophosphatasia (ICD-10: E83.39)
- Only Quest patients were included in the analysis

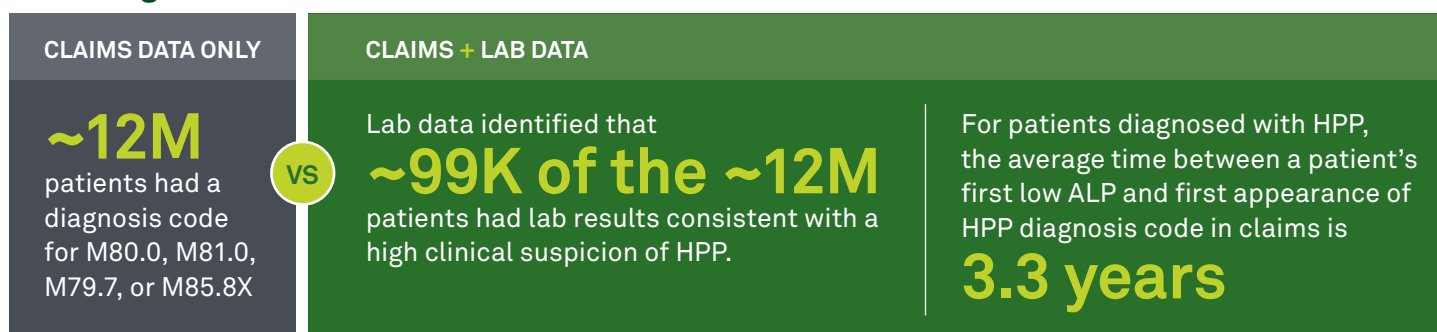
Analysis time frame:

January 2020–December 2025

Clinical results with strong indication of HPP:

2 or more low alkaline phosphatase results (ALP < 35 U/L)

Data insights



Pharma commercial teams can leverage these lab insights to



Drive high-precision HCP targeting

By identifying the ~99K patients with clinical results indicating HPP, pharma teams can minimize wasted effort, maximize efficiency, and prioritize outreach to high-value HCPs actively managing suspected HPP patients.



Accelerate time-to-treatment

By intervening during the typical 3.3-year diagnostic delay, lab data helps equip sales teams to proactively educate prioritized physicians on the correct diagnostic pathway, prompt necessary confirmatory testing, and position their targeted therapy.