

Case 3 (January 22 2009): [N Engl J Med. 360\(4\):398-407](#)

Demographic: Male, 9 months, America North, General speciality

Clinical features extracted from the case and entered into the query box:

bulging fontanelle
nasal congestion
fever
watery diarrhea
seizures
apnea
hypocalcemia
hypophosphatemia
elevated alkaline phosphatase level
anemia

Synonyms used by Isabel for the above query:

apnea
hypocalcemia
nasal congestion
seizures
watery diarrhea
anemia
fever
bulging fontanelle
hypophosphatemia

STOP !

Before you read further you might want to construct your own:

- Complete differential diagnosis
- Final diagnosis

In the following section you will find:

- The differential diagnosis constructed by the MGH panel
- The final diagnosis of the case
- Which of the MGH panel differential diagnoses did Isabel contain

- Did Isabel suggest the Final diagnosis

Differential Diagnoses considered by the MGH panel:

Meningitis

Final Diagnosis of the case according to NEJM:

Rickets due to Vitamin D Deficiency

Differential Diagnoses of the case as given by Isabel:

Meningitis – under Infectious diseases heading

Was the final diagnosis given by Isabel:

Yes, Rickets under Orthopedic disorders

Isabel Differential:

<p>+ INFECTIOUS DISEASES << click to expand</p> <p><input type="checkbox"/> Influenza and Parainfluenza Viruses ? RD</p> <p><input type="checkbox"/> Bacterial Meningitis ? RD</p> <p style="padding-left: 20px;">Streptococcus Pneumonia Infection ? RD</p>	<p>+ ORTHOPEDIC DISORDERS << click to expand</p> <p><input checked="" type="checkbox"/> Vit D Deficient Rickets / Osteomalacia RD</p> <div style="border: 1px solid black; background-color: #fff9c4; padding: 5px; margin: 5px 0;"> <p>Why did this diagnosis come up ?</p> <p>We matched the terms: hypocalcaemia fits anaemia anaemic hypophosphataemia hypophosphate alkaline alkaline phosphatas phosphatase level</p> <p>Degree of match between query entered and isabel database (Not clinical probability): 60%</p> </div> <p>Osteopetrosis ? RD</p> <p>Vit D-Dependent Rickets ? RD</p> <p>X-linked Hypophosphatemic Rickets ? RD</p>
<p>+ NERVOUS SYSTEM DISORDERS << click to expand</p> <p><input type="checkbox"/> Epilepsy & Seizure Disorders ? RD</p>	<p>+ ENDOCRINE SYSTEM << click to expand</p> <p><input type="checkbox"/> Hyperthyroidism ? RD</p>
<p>+ NEPHROLOGY << click to expand</p> <p><input type="checkbox"/> Renal Failure ? RD</p>	<p>+ NEOPLASTIC DISEASES << click to expand</p>
<p>+ LIVER DISORDERS << click to expand</p>	<p>+ METABOLIC DISEASES << click to expand</p>
<p>+ GASTROINTESTINAL DISORDERS << click to expand</p>	<p>+ RESPIRATORY DISORDERS << click to expand</p>
<p>+ TOXICOLOGY << click to expand</p>	<p>+ IMMUNOLOGIC SYSTEM << click to expand</p>
<p>+ NUTRITIONAL DISORDERS << click to expand</p>	<p>+ DERMATOLOGY << click to expand</p>
<p>+ TRAUMA INJURIES << click to expand</p>	