

Case 20 (June 26 2008): [N Engl J Med. 358\(26\):2813-2825](#)

Demographic: Female, 57 yrs, Not-pregnant, America North, General speciality

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

recurrent lower abdominal pain
pelvic pain
red urine
hyponatremia
confusion
seizures
progressive motor weakness
tachycardia
hypertension
constipation
hyperpigmentation

Synonyms used by Isabel for the above query:

Pelvic pain
Hyponatremia
Confusion
Seizures
Tachycardia
Hypertension
Constipation
Red urine

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:

Porphyria – acute porphyria
primarily axonal polyneuropathy
primary demyelinating neuropathy

Final Diagnosis of the case according to NEJM:

Acute porphyria (variegate porphyria).

Differential Diagnoses of the cases given by Isabel:

Acute porphyria – Variegate Porhyria– under Meatabolic diseases heading

Was the final diagnosis given by Isabel:

Yes, under Metabolic diseases heading

Isabel Differential:

+ INFECTIOUS DISEASES << click to expand <input checked="" type="checkbox"/> Measles Infection ? RD	+ RHEUMATIC DISEASES << click to expand <input checked="" type="checkbox"/> Systemic Sclerosis ? RD
+ NERVOUS SYSTEM DISORDERS << click to expand <input checked="" type="checkbox"/> Cerebral Sinus Venous Thrombosis ? RD <input checked="" type="checkbox"/> Multiple Sclerosis ? RD	+ RESPIRATORY DISORDERS << click to expand <input checked="" type="checkbox"/> Bacterial Pneumonia ? RD <input checked="" type="checkbox"/> Pulmonary Thromboembolism ? RD
+ TOXICOLOGY << click to expand <input checked="" type="checkbox"/> Radioactive Isotopes ? RD	+ ENDOCRINE SYSTEM << click to expand <input checked="" type="checkbox"/> Diabetic Ketoacidosis ? RD
+ METABOLIC DISEASES << click to expand <input checked="" type="checkbox"/> Acute Porphyrria ? RD	+ NEPHROLOGY << click to expand <input checked="" type="checkbox"/> Renal Failure ? RD
<div style="border: 1px solid black; background-color: #ffff00; padding: 5px;"> <p>Why did this diagnosis come up ? ✕</p> <p>We matched the terms: hyponatremia low sodium hyponatremic hypertension htn seizures seizure delirious encephalopathy confusion loc lower disorientation constipation tachycardia tachycardic red urine reddish urine abdomen abdominal pain progress progressive motor weak weakness hyperpigment hyperpigmentation</p> <hr/> <p>Degree of match between query entered and isabel database (Not clinical probability): 64%</p> <p>Variegate Porphyrria</p> </div>	+ DERMATOLOGY << click to expand + GYNECOLOGICAL DISORDERS << click to expand + NEUROMUSCULAR DISORDERS << click to expand
+ CARDIAC DISORDERS << click to expand	
+ GASTROINTESTINAL DISORDERS << click to expand	
+ ENVIRONMENTAL HEALTH HAZARDS << click to expand	
+ NUTRITIONAL DISORDERS << click to expand	