

**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:

<b>+ INFECTIOUS DISEASES</b> << click to expand <input checked="" type="checkbox"/> <a href="#">Measles Infection</a> ? RD	<b>+ RHEUMATIC DISEASES</b> << click to expand <input checked="" type="checkbox"/> <a href="#">Systemic Sclerosis</a> ? RD
<b>+ NERVOUS SYSTEM DISORDERS</b> << click to expand <input checked="" type="checkbox"/> <a href="#">Cerebral Sinus Venous Thrombosis</a> ? RD <input checked="" type="checkbox"/> <a href="#">Multiple Sclerosis</a> ? RD	<b>+ RESPIRATORY DISORDERS</b> << click to expand <input checked="" type="checkbox"/> <a href="#">Bacterial Pneumonia</a> ? RD <input checked="" type="checkbox"/> <a href="#">Pulmonary Thromboembolism</a> ? RD
<b>+ TOXICOLOGY</b> << click to expand <input checked="" type="checkbox"/> <a href="#">Radioactive Isotopes</a> ? RD	<b>+ ENDOCRINE SYSTEM</b> << click to expand <input checked="" type="checkbox"/> <a href="#">Diabetic Ketoacidosis</a> ? RD
<b>+ METABOLIC DISEASES</b> << click to expand <input checked="" type="checkbox"/> <a href="#">Acute Porphyrria</a> ? RD	<b>+ NEPHROLOGY</b> << click to expand <input checked="" type="checkbox"/> <a href="#">Renal Failure</a> ? RD
<div style="border: 1px solid black; background-color: #ffff00; padding: 5px;"> <p><b>Why did this diagnosis come up ?</b> <span style="float: right;">✕</span></p> <p><b>We matched the terms:</b> 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><b>Degree of match between query entered and isabel database (Not clinical probability):</b> 64%</p> <p><a href="#">Variegate Porphyrria</a></p> </div>	<b>+ DERMATOLOGY</b> << click to expand <b>+ GYNECOLOGICAL DISORDERS</b> << click to expand <b>+ NEUROMUSCULAR DISORDERS</b> << click to expand
<b>+ CARDIAC DISORDERS</b> << click to expand	
<b>+ GASTROINTESTINAL DISORDERS</b> << click to expand	
<b>+ ENVIRONMENTAL HEALTH HAZARDS</b> << click to expand	
<b>+ NUTRITIONAL DISORDERS</b> << click to expand	