

Not Your Ordinary Rash

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CASE DESCRIPTION

A 17-year-old, 6-week pregnant female presented to the emergency department with symptoms of abdominal pain, nausea, and vomiting for 2 days. Although she initially complained of only low right abdominal pain, by the next day, she complained of a generalized abdominal ache. Laboratory values upon admission included a plasma sodium concentration of 118 mmol/L, potassium of 3.0 mmol/L, and chloride of 80 mmol/L. On the third day of admission, she experienced 2 seizures. She denied previous history of seizures or other neurological diseases, but she did have a long history of “eczema-like” rashes on her face, neck, and hands that had been unresponsive to prednisone. A urine drug screen was negative, and physical examination was unremarkable. On review of her chart, it was noted that 6 months before her hospital admission, the patient had consulted a dermatologist who described numerous small eroded papules and scarring on the back of the patient’s hands, neck, and upper chest. Urine and plasma specimens collected during this dermatology appointment had been sent to a reference laboratory for porphyrin fractionation and porphobilinogen (PBG)³ measurement. Results are presented in Table 1. Because of the patient’s symptoms upon presentation to the emergency department and previously increased urine PBG concentration, she was treated with daily hemin infusions for 4 days and her symptoms resolved. Over the consequent 2 days, her plasma sodium concentration increased to 135 mmol/L and she was discharged.

Table 1. Plasma porphyrin fractionation, urine PBG, and urine porphyrin fractionation results.

	Analyte	Value
Plasma	Total porphyrins	2.5 (≤ 1.0 $\mu\text{g/dL}$)
	Uroporphyrin	<0.1 (≤ 1.0 $\mu\text{g/dL}$)
	Heptacarboxyl porphyrins	<0.1 (≤ 1.0 $\mu\text{g/dL}$)
	Hexacarboxyl porphyrins	<0.1 (≤ 1.0 $\mu\text{g/dL}$)
	Pentacarboxyl porphyrins	0.1 (≤ 1.0 $\mu\text{g/dL}$)
	Coproporphyrin	0.2 (≤ 1.0 $\mu\text{g/dL}$)
Urine	Protoporphyrin	2.1 (≤ 1.0 $\mu\text{g/dL}$)
	Porphobilinogen	16.2 (≤ 1.3 $\mu\text{mol/L}$)
	Uroporphyrin	74 (≤ 30 nmol/L)
	Heptacarboxyl porphyrins	18 (≤ 7 nmol/L)
	Hexacarboxyl porphyrins	5 (≤ 2 nmol/L)
	Pentacarboxyl porphyrins	33 (≤ 5 nmol/L)
	Coproporphyrin	424 (≤ 110 nmol/L)

QUESTIONS TO CONSIDER
• What are some causes of concurrent hyponatremia and abdominal pain?
• How would acute porphyria be differentiated from other conditions with a similar clinical presentation?
• Why did the patient's symptoms resolve following hemin infusion?
• How should molecular testing be integrated into the further workup of this patient?

Final Publication and Comments

The final published version with discussion and comments from the experts will appear in the June 2019 issue of *Clinical Chemistry*. To view the case and comments online, go to <http://www.clinchem.org/content/vol65/issue6> and follow the link to the Clinical Case Study and Commentaries.

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