Solving The Diagnostic Puzzle: A Case of Acute Intermittent Porphyria

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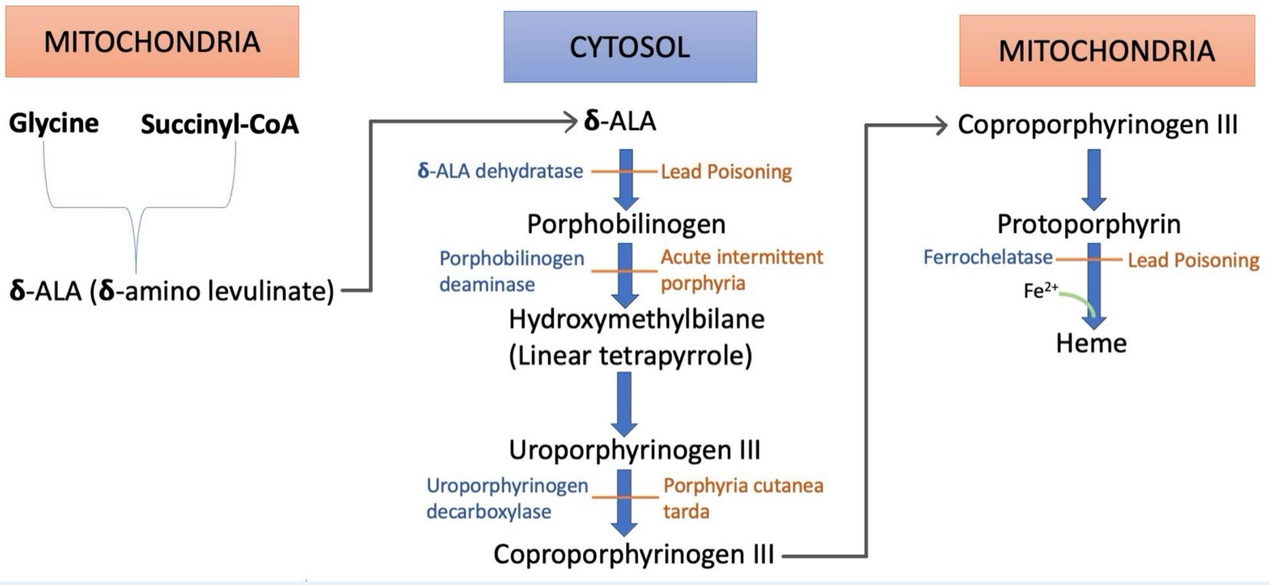
BACKGROUND

Acute intermittent porphyria (AIP) is a rare (50 per million) and debilitating metabolic disorder presenting with unexplained neurovisceral symptoms, making diagnosis challenging. We present a case of a young female who had multiple hospitalizations for severe abdominal pain, ultimately diagnosed with AIP and treated effectively with intravenous hemin.



Urine color change in acute porphyria

CASE PRESENTATION

An 18-year-old woman with vocal cord dysfunction presented with multiple episodes of abdominal pain, nausea, and vomiting over several months. Her physical exam was normal. The liver enzymes were intermittently elevated: AST as high as 46 (8-33 U/L) and ALT as high as 37 (4-36 U/L). ALP and bilirubin were normal. She had mild hyponatremia (132 mEq/L), and lipase was normal. Extensive evaluations, including tissue transglutaminase antibody, CT scan, endoscopy, and colonoscopy, were negative for gallstones, celiac disease, *Helicobacter pylori*, and inflammatory bowel disease. Upon further inquiry, her symptoms coincided with her menstrual cycles. She also experienced episodes of gross hematuria, tingling in her fingertips, and weeks of joint pain, which she attributed to stress from college exams during that time. Urinalysis revealed hematuria, ketonuria, and bilirubinuria. The urine drug screen was normal. Given her recurrent abdominal symptoms with hematuria, porphyria testing was conducted, revealing significantly elevated plasma porphyrin of 20.1 (0.1-1.0 ug/dL) and urine total porphyrin of 15,754 (25-144 nmol/L), highly suggestive of AIP. Genetic testing confirmed a diagnosis of AIP with a pathogenic HMBS gene mutation. She was started on prophylactic hemin during the luteal phase of her menstrual cycle to suppress the overproduction of porphyrin precursors along with symptomatic pain management. She developed homocysteinemia as a side effect of her hemin treatment, which was managed with B-complex vitamins. Over the following months, she experienced fewer and less severe attacks.

CONCLUSION

- This case underscores the need to consider AIP in patients with unexplained recurrent abdominal pain and unexplained neuro-psychiatric symptoms to make a timely diagnosis.
- Further research is needed to optimize AIP's long-term management and explore the full potential of emerging therapies.

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