A Case of Acute Intermittent Porphyria Complicating Pregnancy

Preetpal Grewal, D.O., PGY-III
Claudia Suarez, M.D.
Jose Gonzalez, M.D.
Texas Tech University Health Sciences Center

INTRODUCTION AND BACKGROUND
Acute intermittent porphyria is the most common and most se-
vere of the inherited hepatic porphyrias. This condition is an
autosomal dominant disorder resulting from a partial deficiency
of porphobilinogen deaminase activity, an enzyme in the path-
way of heme synthesis [1, 2]. It is also called Swedish porphyria,
pyrroloporphyria, or intermittent acute porphyria. The estimated
prevalence of the disorder is 5-10 case per 100,000 [1, 2]. The
usual clinical presentation involves abdominal pain, gastrointestinal
symptoms and autonomic nervous system disturbances.

CASE PRESENTATION
This is a 27 year old G3P2 who began prenatal care in our high
risk obstetrics clinic at 23 weeks gestation. She had history of
acute intermittent porphyria diagnosed during the previous year.
The patient was first admitted to our service with complaints of
severe abdominal pain, nausea and several episodes of vomiting.
She also complained of pain in her back and legs. She had
experienced similar symptoms at 20 weeks gestation. Her history
included an admission around 7 weeks gestation for being in a
coma. It was at that time she was found to be pregnant. Since her
initial admission, the patient had recurrent attacks of abdominal
pain and nausea requiring multiple hospital admissions. Her treat-
ment consisted of a high carbohydrate diet of more than 400 gm/
day and narcotics for pain control. Additionally, to prevent other
attacks, she was advised to avoid prolonged periods of fasting.
She incidentally developed mild hypochromic, microcytic ane-
mia that was treated. Her laboratory work up included urine analy-
sis, a glucose tolerance test, and renal function tests, all results
were within normal limits. The pregnancy was monitored closely
for hypertension and fetal growth and any other symptomatic
attacks. The patient delivered a single live born female at 39
weeks with no complications.

DISCUSSION
The prevalence of acute intermittent porphyria varies in the dif-
ferent areas of the world. Though its association with pregnancy
is rare, it presents the obstetricians with challenging problems in
diagnosis and management. Porphyria is difficult to diagnose
during pregnancy because the symptoms simulate hyperemesis gravidarum or eclampsia. It is thought to be associated with dis-
mal maternal and perinatal outcome. During the acute attack, the
neurological dysfunction can involve any portion of the ner-
vous system. An imbalance in the autonomic innervation of the
intestine leads to abdominal pain. Peripheral neuropathy such as
pain in the back and legs is always preceded by abdominal pain.
Other autonomic neuropathies like sweating, vascular spasm,
labile hypertension, and sinus tachycardia may occur. More se-
vere symptoms involve the development of respiratory paraly-
sis, central nervous dysfunction along with seizures, coma, hy-
pothalamic dysfunction, and disruptions with cerebellar and basal
ganglion function [3]. However, recent reports are more encour-
aging due to better understanding of the disease process and
improved prenatal care [4, 5]. Another pregnancy problem is the
use of narcotics to control the acute symptoms and the possibil-
ity of opioid withdrawal in the neonate.

Heme synthesis and the porphyrias Schematic representation of heme synthesis and the enzymatic defects in the porphyrias, beginning with the formation of ALA from succinyl CoA and glycine. The enzymatic defects in each porphyria is shown by a red broken line. In patients, the substrate for the defective enzyme accumulates and is secreted into urine or stool. ALA = alpha-aminolevulinic acid; PBG = porphobilinogen; HPB = hydroxymethylbilane; Uroporphyrinogen = coproporphyrinogen; Protoporphyrinogen = uroporphyrinogen; ILSA = L-10-keto-dehydrobiliverdin; ALA synthase = ALA dehydratase porphyria; AIP = acute intermittent porphyria; COP = congenital erythropoietic porphyria; PCT = porphyria cutanea tarda; HEP = hereditary erythropoietic porphyria; HCP = hereditary coproporphyria; VP = variagated porphyria; EPP = erythropoietic protoporphyria.

Continued on page 15
Females appear to have more severe symptoms and attacks may be precipitated by menstruation, pregnancy, and oral contraceptive use. Normal pregnancy is associated with urinary excretion of aminolevulinic acid, porphobilinogen, and porphyrinas that is 15-60% greater than in non-pregnant females making the diagnosis of acute intermittent porphyria more difficult [4-6]. Pregnant patients with acute intermittent porphyria have higher incidence of acute attacks of porphyria (24-95%), resulting in considerable perinatal mortality (2-42%), high rates of spontaneous abortion (6-12%), hypertension (16%), and low birth weight infants [5]. Patients are instructed to avoid precipitating factors. A high carbohydrate diet (more than 400 gm/day) can decrease porphyrin precursor excretion [4]. For abdominal pain, phenothiazines can be used. Autonomic manifestations such as hypertension and tachycardia can be controlled with the use of propranolol. In patients who do not have coagulopathies, the use of hematin can be considered. Critical monitoring of vitals, intake, output, fluid and electrolyte balance is essential. Porphyrogenic drugs should be avoided and infections should be treated. All these measures are likely to result in a better pregnancy outcome. The infants should be followed as acute intermittent porphyria is an autosomal dominant condition with variable penetrance.

REFERENCES
1. Sassa S. Acute intermittent porphyria. Up to Date http://www.utdol.com/utd/content/

Preetpal Grewal, D.O., PGY-III, Department of OB/GYN at Texas Tech University Health Sciences Center in El Paso.

Claudia Suarez, M.D., Assistant Professor, Assistant Residency Program Director and Director of the Minimally Invasive Surgical Laboratory in the Department of OB/GYN at Texas Tech University Health Sciences Center in El Paso.

Jose Gonzalez, M.D., Associate Professor, Director of Maternal-Fetal Medicine, Vice-Chair of Clinical Affairs and Residency Program Director in the Department of Obstetrics and Gynecology at Texas Tech University Health Sciences Center in El Paso.