Clinical Case Study

Cirrhosis Originally Diagnosed to Nonalcoholic Steatohepatitis

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CASE DESCRIPTION

A 52-year-old white woman with a presumptive diagnosis of cirrhosis attributed to nonalcoholic steatohepatitis was referred for a liver transplant consultation. The patient reported increased abdominal discomfort, swelling, and lower extremity edema beginning 7 years earlier, as well as intermittent nausea, weakness, and confusion which had increased in frequency during the previous year. While undergoing a cholestectomy for gallstones 2 years previously, the patient was noted to have ascites and a cirrhotic-appearing liver. Results of routine laboratory tests and an abdominal ultrasound performed at that time were consistent with cirrhosis.

The patient, a married housewife with 1 adult son, had a physical appearance that suggested chronic illness but was in no acute distress. She denied any history of drug or alcohol use and reported never having received a blood transfusion. She had no history of childhood liver disease and was unaware of any family history of gastrointestinal malignancy or liver disease. The patient did report, however, that 2 of her 7 siblings died from unknown causes as infants, her father died while in a coma of unknown etiology, her mother died from diabetic complications, and a third sibling died of a gynecologic cancer. The patient's medical history was remarkable for obesity, gastroesophageal reflux disease, recurrent urinary tract infections, and arthritis. Physical examination was unremarkable, with the exception of mildly icteric sclera, spider nevi on the anterior chest wall, and bilateral lower extremity edema. Moderately decreased breath sounds in her lower left lung were also noted. Chest x-rays and spirometry were unremarkable.

Laboratory data were consistent with end-stage liver disease and also revealed mild anemia as well as thrombocytopenia and chronic kidney disease. Although at the time of her referral to our institution the patient had a presumptive diagnosis of cirrhosis related to nonalcoholic steatohepatitis, we investigated other causes for her liver failure. The patient tested positive for hepatitis A IgG and negative for hepatitis A IgM antibodies, indicating a remote infection with hepatitis A virus. Hepatitis B and C virus serological tests were all negative. Serological markers were negative for antinuclear, anti–neutrophil cytoplasmic, antimitochondrial, and anti–smooth muscle antibodies. Hemochromatosis genetic tests by PCR for the C282Y, H63D, and S65C point mutations were negative.

Questions to Consider
• Who should be screened for A1AT deficiency?
• What clinical symptoms are typically associated with A1AT deficiency?
• What is the most severe disease associated phenotype?

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Final Publication and Comments

The final published version with discussion and comments from the experts will appear in the August 2008 issue of *Clinical Chemistry*. To view the case and comments online, go to http://www.clinchem.org/content/vol54/issue8/ and follow the link to the Clinical Case Study and Commentaries.

Educational Centers

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