

Pancytopenia and Peripheral Neuropathy in a Woman with Altered Liver Function Tests

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CASE DESCRIPTION

A 39-year-old woman was referred to the digestive and liver specialist after several weeks of her feeling tired, with complaints of abdominal pain, nausea, and vomiting, and with abnormal liver function tests. The patient was in good general health, with a body mass index of 21.2 kg/m², no relevant medical history, and a normal physical examination. The stigmata of chronic liver disease were absent. The patient reported that she had been consuming 3 to 4 beers/day and was not taking any medication. An abdominal ultrasonography revealed hepatic steatosis. Blood and urine tests showed the following: aspartate transaminase, 95 U/L [reference interval (RI)³, 0–32 U/L]; alanine transaminase, 66 U/L (RI, 0–33 U/L); alkaline phosphatase, 180 U/L (RI, 35–105 U/L); γ -glutamyl transferase, 1005 U/L (RI, 0–40 U/L); albumin, 34 g/L (RI, 35–52 g/L); total bilirubin, 0.8 mg/dL (13.7 mmol/L) [RI, 0.1–1.2 mg/dL (1.7–20.5 mmol/L)]; direct bilirubin, 0.7 mg/dL (12.0 mmol/L) [RI, 0.0–0.3 mg/dL (0.00–5.1 mmol/L)]; total cholesterol, 546 mg/dL (14.1 mmol/L) [upper reference limit (URL), <200 mg/dL (<5.17 mmol/L)]; triglycerides, 2183 mg/dL (24.7 mmol/L) [URL, <150 mg/dL (<1.69 mmol/L)]; ceruloplasmin, <3 mg/dL (RI, 16–45 mg/dL); serum copper, 63 μ g/dL (RI, 80–154 μ g/dL); and urine copper, 17 μ g/24 h (RI, 10–50 μ g/24 h). Laboratory tests for other causes of liver disease, such as viral hepatitis B and C, autoimmune hepatitis, celiac disease (endoscopic biopsy), hemochromatosis [transferrin saturation (%)], and α -1 antitrypsin deficiency (serum protein electrophoresis pattern), were negative. A hepatic biopsy was performed, and histological findings included grade 2 to 3 fibrosis and ductular proliferation, with glycogenated nuclei and cytoplasmic microvacuoles and macrovacuoles in hepatocytes. However, microsteatosis could not be confirmed. A quantitative hepatic copper determination revealed a concentration of >739 μ g/g tissue. Genetic testing for the ATP7B⁴ gene was performed, but no mutations were found.

QUESTIONS TO CONSIDER

- What are the most frequent causes of abnormal liver function tests?
- What are the most common causes of hypertriglyceridemia?
- What are the potential causes of low serum ceruloplasmin concentrations?
- What is the most likely diagnosis?

Final Publication and Comments

The final published version with discussion and comments from the experts will appear in the August 2019 issue of *Clinical Chemistry*. To view the case and comments online, go to <http://www.clinchem.org/content/vol65/issue8> and follow the link to the Clinical Case Study and Commentaries.

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