Alström Syndrome with Acute Pancreatitis: A Case Report

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We report the case of a 21-year-old female with Alström syndrome who also suffered from acute pancreatitis of obscure manifestation. The patient had underlying cone-rod dystrophy of the retinas, nystagmus, obesity, progressive sensorineural hearing impairment, diabetes mellitus, and hypertriglyceridemia, compatible with the clinical diagnosis of Alström syndrome. Serial examinations showed liver dysfunction and pancreatitis. In treating a patient with poor communication (i.e. cone-rod dystrophy and hearing impairment) suffering from acute illness, understanding the underlying disease and the potential for pancreatitis with hypertriglyceridemia is necessary. It is also a challenge to treat a patient with multiple system involvement. In conclusion, Alström syndrome is a disease of systemic multi-organ involvement, and hepatic disease and pancreatitis, possibly due to dyslipidemia, appear to be manifestations of Alström syndrome.

Key Words: Alström syndrome, acute pancreatitis
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Alström syndrome is a rare autosomal recessive disease. Its major clinical features are cone-rod dystrophy of the retinas, resulting in childhood blindness, truncal obesity that manifests during childhood, insulin-resistant diabetes mellitus, progressive sensorineural hearing loss, and infantile- or adolescent-onset dilated cardiomyopathy [1,2]. As the patient suffered from visual and hearing impairment, and problems associated with mental retardation, further laboratory investigations to determine if other organ systems were involved, an understanding of the syndrome and a high index of suspicion of other specific diseases were required. Hyperinsulinemia and hypertriglyceridemia are frequent in childhood. Children with dyslipidemia are at risk of pancreatitis. We report the case of a 21-year-old patient with Alström syndrome who suffered from pancreatitis with obscure symptoms and signs.

Case Presentation

The patient was a 21-year-old woman with Alström syndrome manifested by cone-rod dystrophy and nystagmus, obesity, progressive sensorineural hearing loss, diabetes mellitus, and hypertriglyceridemia. Her parents were healthy but she had a younger brother who also had Alström syndrome. She was intermittently monitored at our hospital and glibenclamide and gemfibrozil had been prescribed.

She was sent to our hospital due to dull abdominal pain. Physical examination showed pale conjunctivae

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and dull abdominal pain without rebounding pain. Her blood glucose level was 439 mg/dL. Blood osmolarity was within the normal range. Arterial blood gas analysis showed mild metabolic acidosis, with a base excess –5.7 mEq/L. Routine urinalysis was negative for ketones, proteinuria, and pyuria. Thus, diabetic ketoacidosis and nonketotic hyperosmolar syndrome, acute complications of diabetes mellitus, were eliminated.

A complete blood count showed a hematocrit of 37.4 g/dL, a white blood cell count of 34,380/μL, and a normal platelet count. Blood urea nitrogen was 29 mg/dL, serum creatinine was 1.68 mg/dL, and albumin level was normal. Alanine aminotransferase (ALT) was 72 U/L (normal range, 10–40 U/L), aspartate aminotransferase (AST) was 54 U/L (normal range, 10–35 U/L), γ-glutamyltransferase (GGT) was 126 U/L (normal range, 0–48 U/L), lactate dehydrogenase was 434 U/L (normal range, 90–181 U/L), and serum total calcium was normal. Hb_A1_C was 11.0% (normal range, < 6%). Triglycerides were 44.4 g/L (normal range, 3.5–15.3 g/L), total cholesterol was 23.1 g/L (normal range, 12.5–20.0 g/L), serum amylase was 323 U/L (normal range, 63–123 U/L), and serum lipase was 1,530 U/L (normal range, 7–58 U/L).

The diagnosis of acute pancreatitis was suspected and abdominal computed tomography (CT) was performed (Figures 1 and 2), which showed swelling of the pancreas and fluid accumulation in the right perinephric area, hepatomegaly, splenomegaly, and distension of the stomach. CT grading according to Balthazar et al [3] was C. Ranson's criteria score was 4. The patient was diagnosed with severe acute pancreatitis.

Echocardiography was performed to assess the possibility of dilated cardiomyopathy. The result was negative. The follow-up CT revealed resolution of fluid accumulation in the right perinephric area. A liver biopsy was performed to determine the etiology of hepatomegaly and showed mild hepatic fibrosis, periportal inflammation, and fatty liver. After intensive care, the patient's condition stabilized and she was discharged 20 days later.

Discussion

We report a rare case of Alström syndrome complicated by acute pancreatitis in a 21-year-old woman. Alström syndrome is inherited in an autosomal recessive manner. The diagnosis is based on clinical findings. Molecular genetic testing for the *ALMS1* gene (chromosomal locus 2p13) is available on a research basis only [4,5]. About 200 patients with Alström syndrome are known worldwide [6]. The patient's diagnosis at the time of initial evaluation was Alström

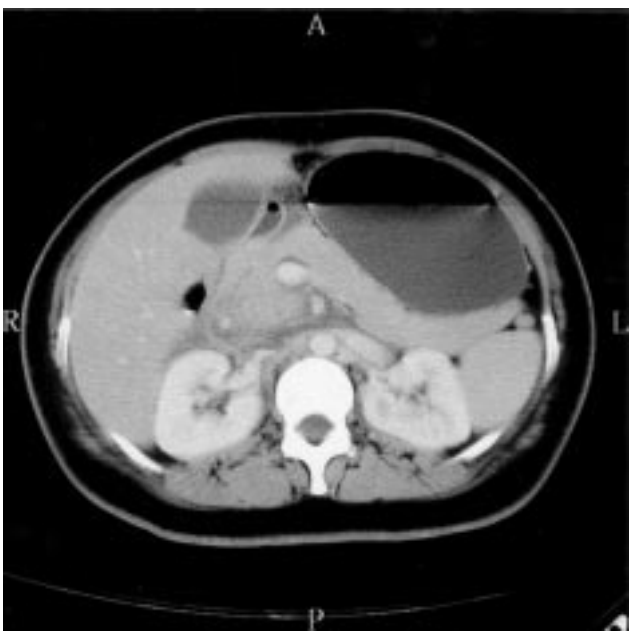

Figure 1. *Abdominal computed tomography with enhancement showed swelling of the pancreas and fluid accumulation in the right perinephric area, which are compatible with pancreatitis.*

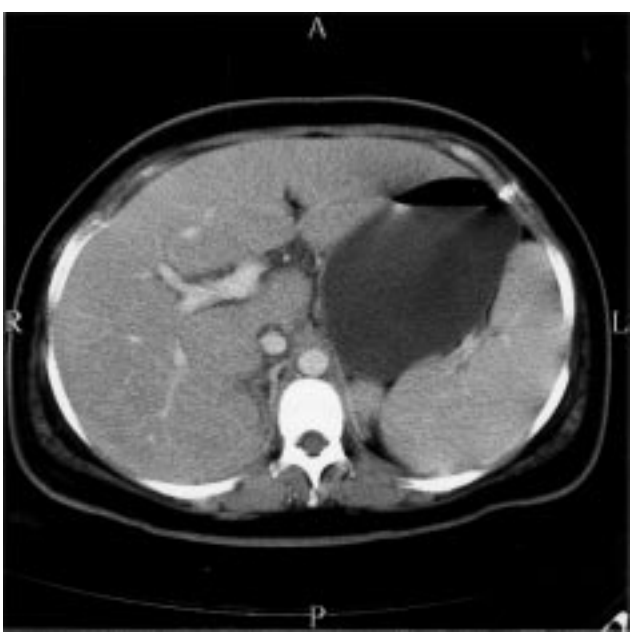

Figure 2. *Abdominal computed tomography with enhancement showed hepatomegaly, splenomegaly, and stomach distension.*

syndrome; a detailed history of the disease helps to direct a detailed physical examination and selected investigations. Long-term management depends on the organ systems involved.

Abnormal liver function is common in early childhood and can progress to hepatic failure in the second to third decades of life and is common in Alström syndrome. Measurement of plasma ALT, AST, and GGT concentrations should be performed yearly. The presence of high plasma ALT, AST, and GGT concentrations is an indication for liver ultrasonography to evaluate possible hepatomegaly and portal hypertension. Our patient showed hepatic dysfunction, hepatomegaly, and splenomegaly. Hepatic dysfunction appeared to be a manifestation of Alström syndrome. Liver biopsies and postmortem examinations have revealed varying degrees of hepatic fibrosis, cirrhosis, and fatty liver [7,8]. The liver biopsy of our patient showed similar results.

Serum triglyceride and cholesterol concentrations should be measured annually or when the patient is ill and/or dehydrated, because pancreatitis, precipitated by hyperlipidemia, can be life-threatening. The patient's initial cholesterol and triglyceride levels were 25.0–30.0 g/L and 30.0–35.0 g/L, respectively. Our patient had hypertriglyceridemia, thus confirming the diagnosis of pancreatitis, although the presenting symptoms were obscure.

In conclusion, Alström syndrome is a systemic disease of multiple organ systems. Hepatic disease and pancreatitis, possibly due to dyslipidemia, appear to be manifestations of Alström syndrome.

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