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Case Report

Acute Pancreatitis: a Rare Complication in a Patient with Senior Loken Syndrome

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Abstract

Introduction: Senior-Loken syndrome is a rare entity that combines familial nephronophthisisand retinal dystrophy. It has an autosomal recessive inheritance pattern and is characterized by a chronic tubulointerstitial nephritis that progress to terminal renal failure during the first or second decades of life. Systemic associations of this syndrome include sensorineural hearing loss, liver fibrosis or cerebral vermis hypoplasia. Acute pancreatitis has not been previously reported in this syndrome.

Case report: This is a 28- years-old patient who was diagnosed to have Senior Loken syndrome at the age of 10 years because of renal impairment and tapetoretinal degeneration and was later started on regular hemodialysis. He had no family history of renal disease, hypertriglyceridemia or cholelithiasis. He presented to our center complaining of acute abdominal pain and vomiting. He had abdominal tenderness without guarding. Investigations revealed a lipase level of 3856 IU/l and an abdominal CT scan showed features of acute pancreatitis. The abdominal ultrasound showed no biliary tree malformations or gallstone obstruction. He had no history of recent drug intake or alcohol consumption and his serum triglyceride level was normal. A diagnosis of moderate acute pancreatitis was made and the patient was managed conservatively with good outcome.

Conclusion: Taking into consideration the uncertainty about the presence of liver fibrosis and the fact that imaging may have missed a passing gallstone, this case may indicate another rare systemic complication of Senior-Loken syndrome.

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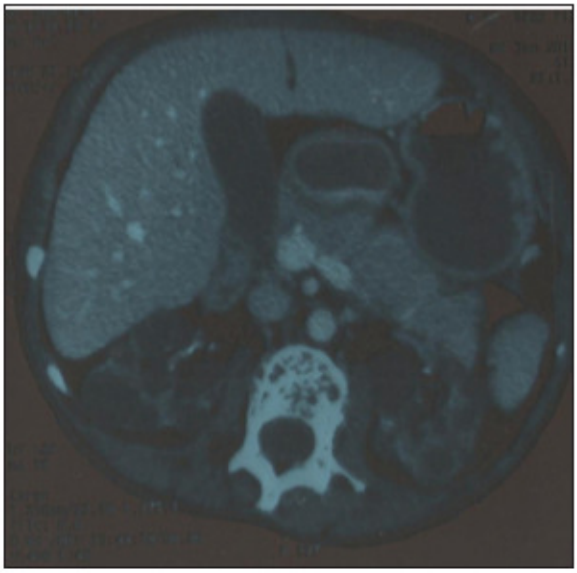
Introduction

Senior-Loken syndrome is a rare syndrome that combines familial juvenile nephronophthisis and retinopathy. Senior et al first described it in 1961[1], and in the same year, Loken et al [2] described the same condition in two siblings. It has an autosomal recessive inheritance pattern and is characterized by a chronic tubulointerstitial nephritis that progress to terminal renal failure during the second decade (juvenile form) or before the age of 5 years (infantile form). The earliest presenting features include an impaired urinary concentrating ability leading to polyuria and polydipsia. The clinical features of this syndrome may include systemic involvement in the form of sensorineural hearing loss, liver fibrosis or cerebral vermis hypoplasia [3]. To the best of our knowledge, this is the first report of acute pancreatitis in a patient with Senior-Loken syndrome.

Case Report

This is a 28-years-old patient born to nonconsanguineous parents. There was neither any family history of renal diseases, nor hearing or visual impairment. He was referred to our center at the age of 10 years because of renal failure. He was relatively well until he developed symptoms of polyuria and polydipsia during his childhood period. Abdominal ultrasonography showed bilateral normal-sized kidney with hyperechogenic cortex and a medullary cyst in the right kidney. His vision decreased gradually, becoming especially impaired at night. He was blind by the age of 12. On ophthalmologic evaluation, tapetoretinal degeneration was detected. Based on the clinical picture; the early childhood blindness and nephronophthisis our patient was diagnosed as having the Senior-Loken syndrome. Unfortunately, family work-up, genetic testing and liver biopsywere not performed.

Figure1: Abdominal CT scan showing pancreatic necrosis. Note normal seized kidneys with multiple cysts



The patient was discharged on conservative management for his chronic kidney disease and follow-up was arranged in a nephrology unit. He was subsequently started on hemodialysis once he reached end stage renal disease (ESRD). A living donor was not available and he was enrolled in the waiting list for renal transplantation.

He was admitted in our center at the age of 28 years for the evaluation of acute abdominal pain and vomiting. No personal history of drug intake or alcohol consumption was reported. Furthermore, the patient had no family history of hypertriglyceridemia or cholelithiasis. On examination the patient was found to be pale. Abdominal tenderness in the epigastric region was demonstrated without guarding. He was neither jaundiced nor edematous. He was blind and had nystagmus. The growth parameters were under the 25th centile. He did not have any dysmorphic features. The laboratory investigations were consistent with the diagnosis of acute pancreatitis. Lipase level was 3856 IU/l (48 times the high normal value). Hemoglobin was 9 g/dl with normal leukocyte and platelet counts. Blood urea nitrogen was 49.6 mg/ dL and serum creatinine was 9.2 mg/dL. Serum sodium and potassium levels were normal. Serum triglycerides and cholesterol levels were normal at 143 mg/ dL and 170 mg/ dL; respectively. Abdominal CT scan showed acute pancreatitis, Balthazar Stage-E, with a modified CT severity index of the American Roentgen Ray Society of 6 (Figure-1). The abdominal ultrasound showed no biliary tree abnormalities or gallstone obstruction. Basedon these findings the diagnosis of moderate acute pancreatitis was entertained.

The patient was kept nil per mouth while maintained on intravenous fluid hydration for three days. Analgesics were administrated for abdominal pain relief. The evolution was favorable, enteral feeding was allowed on day-3 with good tolerance. No complications were noted and antibiotic therapy was felt unnecessary. Follow up abdominal CT was normal.

Discussion

The association of nephronophthisis and tapeto-retinal degeneration occurs in approximately 10-15% of all the cases of nephronophthisis. Besides renal impairment and retinal degeneration, the clinical associations of Senior Loken syndrome include sensorineural hearing loss, liver fibrosis or cerebral vermis hypoplasia. The ocular involvement takes several forms, it could manifest with early and severe congenital amaurosis of the Leber type or late-onset pigmentary retinal degeneration which is characterized by night blindness and is followed later by complete visual loss. The tapeto-retinal degeneration varies in its nature and severity. It is characterized by a progressive degeneration of the choroid and the retina with an early and complete extinction of the electro-retinogram [4]. Other ocular findings include cataract, Coat's disease and keratoconus [5]. The ophthalmic findings in our patient were important clues that led to the diagnosis of Senior-Loken syndrome. A careful ophthalmic examination was helpful in correctly diagnosing the syndrome and should be practiced in all young patients with renal failure. In identified cases, an annual eye examination which commences at the time of the diagnosis is recommended. An electro-retinogram helps in the diagnosis of theses varieties before the observation of retinitis pigmentosa on fundoscopic examination.

The renal involvement of this syndrome is well defined [2]. The features as well as the clinical course are identical to those of isolated nephronophthisis which is usually insidious in nature and progressive reaching end stage renal failure before the age of 20 years. However, late onset renal failure in the third and fourth decades has been reported [6]. Polyuria, polydipsia and impaired concentrating ability are the earliest clinical signs. In Senior-Loken syndrome, proteinuria is inconsistent and urinary sediment is usually normal. Hematuria is not a feature and blood pressure remains normal until chronic kidney disease develops. Tubular lesions predominate with defective concentrating ability as an invariable sign. Urinary acidification ability is usually preserved but hyperchloremic acidosis may occur and serum electrolytes may be disturbed with hypernatriuesis [2, 6].

The main histological findings in kidney biopsy are the presence of tubulointerstitial fibrosis, tubular dilatation and tubular atrophy, with thickening and multi-layering of the tubular basement membrane. The glomeruli are basically normal, but a secondary glomerosclerosis can be seen in advanced nephronophthisis [7]. To date, there are no proven treatments for nephronophtisis. At present, management is mainly directed to delaying the progression of the renal failure which leads to end stage renal disease and the need for dialysis and transplantation.

When diagnosing acute pancreatitis, attempts to determine its underlying etiology are undertaken. In particular those causes that may affect acute management should be carefully sought. Relevant historical clues include any previous diagnosis of biliary tract disease or gallstones, cholecystectomy, other biliary or pancreatic surgery, acute or chronic pancreatitis or their complications. Enquiries should be made regarding use of alcohol, medications and the timing of their initiation, recent abdominal trauma, weight loss or other symptoms suggesting a malignancy, or a family history of pancreatitis. This systematic approach was adopted in our patient, and it had shown no underlying clear etiology for his acute pancreatitis episode.

Conclusion

Taking into consideration the uncertainty about the presence of liver fibrosis and the fact that imaging may have missed a passing gallstone, this case may indicate another rare systemic complication of Senior-Loken syndrome.

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