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Case Report

An Interesting Case of Pruritus

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Abstract

Progressive familial intrahepatic cholestasis (PFIC) is a group of autosomal recessive inherited disorders which may lead to cirrhosis and its complications. It is rare with an incidence of 1;50000 births. Here, we report a case of PFIC in a young gentleman who presented with cholestastic features. Diagnosis is usually by biopsy and genetic studies. Medical treatment involves UDCA and pruritus relieving measures. Surgery involves biliary drainage and liver transplantation.

Keywords: Pruritus; Jaundice; Cholestasis; Autosomal Recessive

Introduction

Progressive familial intrahepatic cholestasis (PFIC) is an autosomal recessive disorder [1] which presents with jaundice and pruritus. It usually progress to liver cirrhosis. The key is detecting the disease early and institution of appropriate management. Most of the cases usually require surgical management in the form of biliary drainage or liver transplantation. Here, we report a case of PFIC who is responding to medical management and is kept under follow up.

Case Report

32 year old male who is an asthmatic controlled on inhalers, came to gastroenterology OPD with yellowish discoloration of eyes since 2weeks. It was associated with severe pruritus. No history of abdominal pain or weight loss. He gives history of fever 1month back which lasted for 1week and he had taken complementary and alternative medicines for the same. He had recurrent episodes of jaundice lasting 1-2 months since he was 6months of age. Examination revealed deep icterus, no KF ring, mild hepatomegaly.

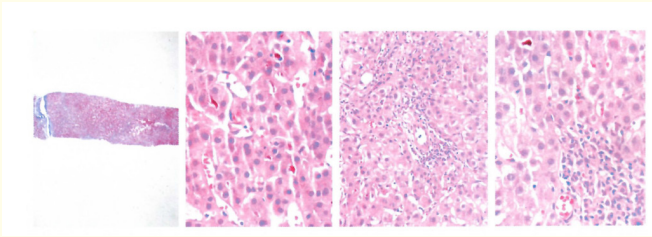
- HB: 13.9 g/dL TLC: 8800/mm^3 PMN: 80% Lymphocytes: 15% Eosinophils: 5%, ESR: 40mm/h, Absolute eosinophil count – 440/mm^3
- Bilirubin (Total)- 12 mg/dL; direct- 5mg/dL, SGOT- 66 IU/L, SGPT-50 IU/L, ALP- 170 U/L(normal <115), protein- 6.8 g/dL, Albumin-3.6 g/dL, GGT- 62 U/L(normal <72), INR-1.1
- Calcium – 9 mg/dL, LDH- 120 U//L

- HBsAg: Negative, Anti HCV: negative, TSH- 2.2 mIU/L, ANA-negative, Total IgG- 85 g/dL
- AMA-M2-negative, ACE level- 44 IU/L.

Chest Xray did not show any infiltrates.

USG abdomen showed hepatomegaly with liver span of 17cm. No IHBRD. Fibroscan- 17.8 kPa. Upper GI endoscopy did not reveal any varices.

A diagnosis of intrahepatic cholestasis with superimposed drug induced liver injury(DILI) was considered. Liver biopsy was performed.



Figure

Liver biopsy revealed maintained acinar architecture with increased periportal, perivenular and perisinusoidal fibrosis. Portal tracts show mixed lymphocytic, eosinophilic and PMN infiltrates. Bile duct appears damaged, atrophic and absent in few of the smaller tracts. Hepatocellular and canalicular cholestasis along with focal mild feathery degeneration, cholestatic rosettes and minimal macrovesicular steatosis. Foci of lobular inflammation seen. No copper associated protein deposits.

IHC for MDR-3 was normal.

Impression

Chronic cholestastic pathology with increased fibrosis. Possibility of PFIC-1 with superimposed drug toxicity to be considered.

Since GGT was normal and with a histopathology report showing increased fibrosis, a diagnosis of PFIC-1 with superimposed DILI was made. He was started on UDCA, N acetyl cysteine, cholestyramine and Vitamin D. His jaundice and pruritus improved and bilirubin came back to normal. He is kept under maintenance dose of UDCA and is on 6 monthly follow up.

Discussion and Conclusion

Progressive familial intrahepatic cholestasis (PFIC) is a group of inherited disorder which usually begins in childhood and later develops into cirrhosis and its complications [1]. It is autosomal recessive. Primarily, 3 types are recognized: PFIC-1, 2 and 3. PFIC-1 is due to a mutation in ATP8B1 gene, PFIC-2 is due to defect in BSEP, PFIC-3 due to defect in MDR3 [2]. Other subtypes with mutation in tight junction protein-2 and FXR have also been recognized [3]. Benign recurrent intrahepatic cholestasis presents in a similar way with intermittent episodes of cholestasis, except that it is primarily benign, without fibrosis in liver biopsy and does not lead to cirrhosis. It is predominantly of 2 types- BRIC-1 and 2. Gene mutations are similar to PFIC 1 and 2 except that it results in partial impairment of protein function in BRIC while it is severe impairment in PFIC [4]. Incidence of PFIC is 1 in 50000 to 100000 births [5]. Extrahepatic manifestations in PFIC -1 includes diarrhea, pancreatitis, wheezing episodes, hearing loss [1]. PFIC-2 is the most aggressive subtype and is usually associated with gallstones and gallbladder malignancies [1]. PFIC-1 and PFIC-2 presented with low GGT, while PFIC-3 had elevated GGT levels. Our patient had intermittent episodes of cholestasis with low GGT finally resulting in liver fibrosis with prior history of wheezing episodes and biopsy suggestive of PFIC. Medical management usually involves UDCA, antipruritic medications and in severe cases biliary drainage [6]. Advanced cases may require liver transplantation.

Conclusion

PFIC is a rare condition. The diagnosis of PFIC should be kept in mind while evaluating a case of intrahepatic cholestasis. Early diagnosis and prompt treatment may be lifesaving.

Statement of Ethics

The patient gave written informed consent to write this case, and the protocol was approved by the institute's committee on human research.

Conflict of Interest Statement

All authors have no financial disclosure or conflicts of interest.

Author Contributions

Dr Neeraj Vinayakumar : Data collection, Manuscript preparation and literature search; Dr.Meenakshi C Nayanar: literature search; Dr K R Vinayakumar: Data interpretation and Manuscript preparation.

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