A Rare Case of Familial Hypercholanemia

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Abstract

Introduction

Sodium taurocholate cotransporting polypeptide (NTCP) is a carrier protein that is encoded by the gene SLC10A1. This protein is expressed in hepatocytes and functions to uptake bile acids from the plasma. Bile acids contribute to many metabolic and hormonal pathways. In the liver, bile acids are synthesized from cholesterol. The NTCP protein is exclusively expressed in the liver and plays an important role in the transport of conjugated bile acids from the portal blood into the liver. This report describes an African-American infant patient with NTCP deficiency, who presented with poor weight gain and jaundice.

Case Presentation

The patient is a 6 week old African American Infant who was referred to the emergency department for further evaluation of poor weight gain and jaundice. Initial lab studies revealed elevated liver enzymes and direct hyperbilirubinemia. The hospital stay was complicated by recurrent bouts of hypoglycemia. Right upper quadrant ultrasound, MRCP, and fluoroscopic guided cholangiogram with liver biopsy were unrevealing. The infant continued to gain weight through her hospital stay and her liver enzymes trended down. She was discharged while liver biopsy results were pending.

Final Working Diagnosis

The pathology report from her liver biopsy showed “diffuse cellular and canalicular cholestasis, some ballooning of hepatocytes and focal giant cell transformation, small interlobular bile ducts, and extramedullary hematopoiesis.” A cholestasis panel was sent to Emory University. Her panel results revealed heterozygosity for familial hypercholanemia — a very rare genetic disorder characterized by elevated serum bile acids, pruritus, and fat malabsorption.

Management/Outcome/Follow Up

Management of NTCP deficiency is primarily supportive due to the relatively mild clinical course, including physical therapy and dietary management. Patients are particularly at risk for growth delay and fat soluble vitamin deficiency. Clinical management should include regular monitoring of liver enzymes.

Learning Objectives

1. Understand physiology of unusual genetic causes of elevated bilirubin in infants
2. Identify presentation and diagnostic workup of causes of elevated bilirubin in infants