Case Description

- IM was a healthy 4 week old M with no relevant family or birth history who presented to the ER with scleral icterus and jaundice, found to have direct hyperbilirubinemia, in addition to transaminits, eosinophilia, in the setting of abnormal facies, murmur, and low weight.
- Lab results: total bilirubin spiked at day 1 of admission at 13.8 mg/dl shown in figure 3, direct bilirubin spiked at day 14 at 10.2 mg/dl shown in figure 4; AST spiked around day 14 at 443 U/L and ALT spiked around day 14 at 244 U/L, both trended down; urine organic acids: indicative of liver disease; bile acids: elevated but inconclusive; genetic cholestasis panel: variant of unknown significance
- Liver biopsy: pathology consistent with PFIC

Introduction

This case follows an infant who initially presented to the ER with scleral icterus and jaundice. The patient also had abnormal physical exam findings. This prompted a further evaluation of these abnormalities. After a thorough workup with interdisciplinary teamwork an unknown diagnosis was made after extensive rule out.

Objective

- Understand the differential diagnosis for neonatal cholestasis.
- Know the work up required for suspected cholestasis, including needed lab work and imaging.
- Be familiar with the workup and treatment of progressive familial intrahepatic cholestasis (PFIC).

Approach

- Reviewed differential diagnoses for neonatal cholestasis, including infection, biliary atresia, Alagille syndrome, alpha-1 antitrypsin, and PFIC.
- Clinical features: PFIC is a clinical syndrome with features of chronic intrahepatic cholestasis that typically begins in infancy and progresses to biliary cirrhosis and hepatic failure by the first or second decade of life. It is characterized by defective secretion of bile acids.
- Lab work: CMP, Bilirubin panel, GGT, PT/INR, PTT, EBV, CMV, hepatitis panel, genetic tests, urine organic acids, bile acids, alpha-1 antitrypsin phenotype, microarray, genetic cholestasis panel
- Imaging: US liver and gallbladder
- Liver biopsy
- Diagnosis: liver biopsy and genetic testing after thorough lab evaluation.
- Treatments: Only known treatment is liver transplantation.

Discussion/Conclusions

- Progressive Familial Intrahepatic Cholestasis is a syndrome which requires significant workup to rule out other pathologies.
- There are many different subtypes of PFIC, which require genetic testing.
- Liver transplantation is the treatment of choice when patients have failed medical treatment and/or biliary diversion and poor quality of life due to pruritis. Transplantation can be done earlier with excellent long-term survival and quality of life.
- This case demonstrates the large differential diagnoses that should be considered when an infant presents with neonatal cholestasis.
- Patients should have broad laboratory testing in addition to US liver and gallbladder, to rule out biliary atresia, the most common cause of cholestasis in his age group.
- After initial testing, diagnosis of PFIC requires a large multidisciplinary team, including gastroenterologists, hepatologists, and genetic counselors. A liver biopsy may also be required for diagnosis.

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References

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