

Common Variable Immune Deficiency with Atypical Hepatic Disease

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INTRODUCTION

- Common variable immunodeficiency (CVID) is a 1° B-cell disorder characterized by hypogammaglobulinemia
- Immune dysregulation theorized to affect liver
- Primarily elevated ALP, nodular regenerative hyperplasia (NRH), cholestasis, portal HTN
- We present case of CVID with atypical liver involvement on imaging/biopsy revealing focal nodular hyperplasia (FNH)

CASE PRESENTATION

- 24-year-old female with PMH of CVID on intravenous immunoglobulin (IVIG) presented for follow-up of chronically elevated liver enzymes, cholestatic pattern, and intact synthetic function
- Medications include Ursodiol and Multivitamin
- No alcohol, recreational or herbal drugs use
- Denies symptoms at present
- Vitals and physical exam within normal limits

Hepatic Function Panel

Alkaline phosphatase (ALP)	1,050 U/L
Aspartate aminotransferase (AST)	114 U/L
Alanine aminotransferase (ALT)	214 U/L
Total bilirubin	1.0 U/L

Previous Workup

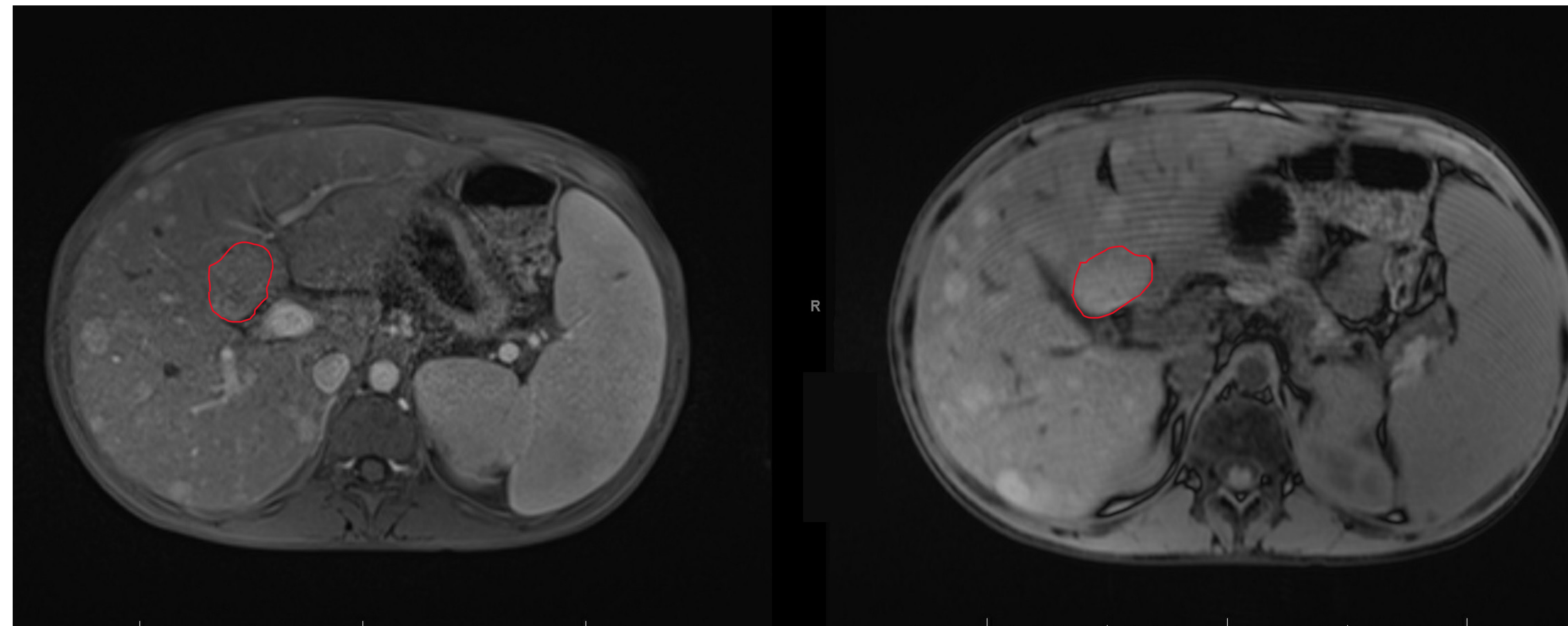
- Antinuclear, anti-smooth muscle, and antimitochondrial antibodies
- Hepatitis A-E serologies, EBV, HSV and VZV PCR
- Genetic testing for Wilson, HH, A1AT

Result

- Negative
- Negative
- Negative

Patient Course

- Persistently elevated hepatic function tests and negative serologic workup prompted Magnetic Resonance Cholangiopancreatography (MRCP) shown below:



- Radiology report read as follows:
 - Normal intra- and extrahepatic biliary tree
 - Innumerable hepatic lesions with characteristics suggestive of multiple focal nodular hyperplasia, largest of which measuring 3.8cm (indicated by the red outlines above)
 - Benign appearing hemangiomas (not visible on above images)

Patient Course (cont.)

- Ultrasound-guided liver biopsy of left hepatic lobe lesion (consistent with FNH) was performed to evaluate cytology
- Two separate pathologists confirmed the radiology findings of FNH
- Patient remains otherwise healthy with stable hepatic function tests

DISCUSSION

- At least 10% of CVID patients present with liver involvement, including anicteric cholestatic liver enzymes (65%) and portal hypertension (50%)
- Histological analysis in recent studies revealed non-fibrosing architectural abnormalities consistent with nodular regenerative hyperplasia in 84% of CVID
- Less commonly, typical histologic features of primary biliary cholangitis.
- Our case liver imaging and biopsy read by two pathologists (at two separate institutions) revealed focal nodular hyperplasia, a new entity not described in previous studies.