



# Rare Case of Liver Fibrosis and Hepatic Manifestations in HNF1B



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## Introduction

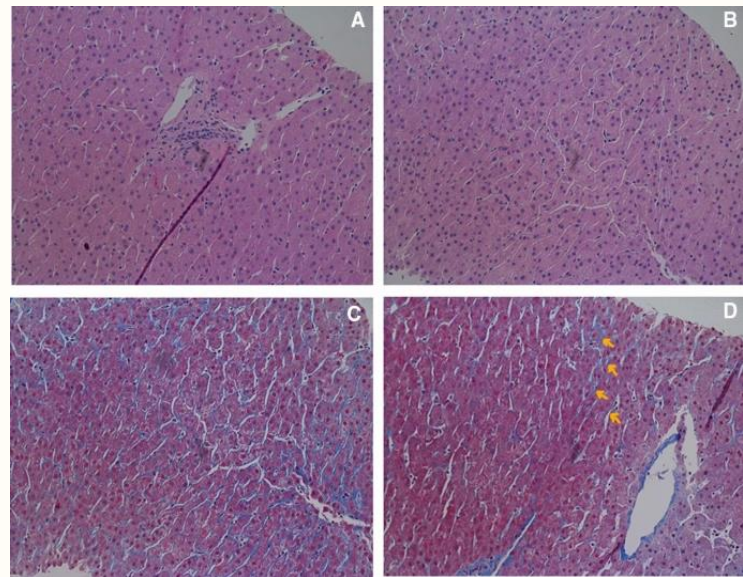
Mature Onset Diabetes of the Young (MODY) is a rare form of DM caused by various mutations. Hepatocyte Nuclear Factor 1-beta (HNF1B) defects are a rare cause of MODY that can result renal cysts and abnormalities of the uterine tract and pancreas. MODY increases the risk of complications of diabetes, such as cirrhosis. Diabetic cirrhosis is often associated with steatosis of the liver. The prevalence of NAFLD in Type 2 diabetics is thought to be 30-50%. Rarely do we see severe liver fibrosis without steatosis. Diabetic hepatosclerosis is thought to be secondary to microangiopathic injury. Such cases present with normal serum aminotransferase levels (ATL) and elevated alkaline phosphatase (AP) levels, likely a result of decreased sinusoidal volume. NAFLD patients present with elevated ATL and normal AP, due to steatohepatitis. We present a case of persistently elevated AP with normal ATL and hepatic fibrosis with biopsy findings of NASH.

## Case Report

A 40-year-old F with a BMI of 20 and PMH of HNF1B mediated MODY, anatomical defects of the pancreas and uterus, and 5 year elevation in AP. Fibroscan confirmed stage 2 fibrosis of the liver and no steatosis. Liver biopsy confirmed Fibroscan results, and showed nodular regenerative hyperplasia (NRH), perisinusoidal fibrosis, and diabetic hepatosclerosis (Figure 1). CT scan of the abdomen showed normal liver and spleen and absent body and tail of pancreas.

Figure 1

Example of Perisinusoidal Fibrosis and Diabetic Hepatosclerosis



Patient Liver Function Test Values

AP	AST	ALT	TB	GGT	Plt	HbA1c	CRP
589	48	75	0.8	662	325	10.0	<0.2

## Discussion

This patient is of particular interest due to their fibrotic liver damage in the presence of diabetes and in the absence of any fatty liver disease. Hepatosclerosis induced liver damage is a rare cause of liver fibrosis in diabetics. NRH is a rare condition that is associated with autoimmune disease, immunodeficiency, hematologic factors, infection, neoplasms, and drug-related cases (2). To our knowledge this is the first published case of HNF1B mediated liver fibrosis and findings of NRH in this patient population. It is important to monitor these patients in regard to progression towards cirrhosis as well as monitoring for presence of non-cirrhotic portal hypertension and its complications seen in patients with NRH.

### References

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- Thung SN, Gerber MA, Bodenheimer HC Jr. Nodular regenerative hyperplasia of the liver in a patient with diabetes mellitus. *Cancer*. 1982 Feb 1;49(3):543-6.