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Liver transplantation in patients with type IIIa glycogen storage disease, cirrhosis and hepatocellular carcinoma

Elena Iglesias-Jorquera[1], Paula Tomás-Pujante[1], Gema Ruiz-García[2], Ángel Manuel Vargas-Acosta[1] and José Antonio Pons-Miñano[1]

Departments of [1]Gastroenterology and Hepatology, and [2]Pathology. Hospital Universitario Virgen de la Arrixaca. Murcia, Spain

Correspondence: Elena Iglesias Jorquera
e-mail: elena_ij@hotmail.com

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Dear Editor,

Type III glycogen storage disease (GSD-III) is an autosomal recessive disorder due to the deficiency of the glycogen debrancher enzyme. 80% of the patients have hepatic and muscular involvement (IIIa), compared to 15% with only liver involvement (IIIb). As the life expectancy improves in these patients, the possible liver complications are better understood.

Case report

We report the case of a male patient diagnosed in childhood with glycogen storage disease (GSD) by a liver biopsy performed via hypertransaminasemia. At 43 years of age, he was referred to our hepatology unit due to chronic liver disease. He suffered

from amyotrophy, global muscle weakness and obstructive hypertrophic cardiomyopathy. The muscle biopsy showed a deficiency of the glycogen debrancher enzyme, compatible with GSD-IIIa.

He progressively developed portal hypertension, with ascites, esophageal varices and gastropathy. At 48 years of age, hepatocellular carcinoma (HCC) of 3.4 cm was diagnosed by ultrasound, abdominal contrast-enhanced computed tomography and magnetic resonance imaging. The multidisciplinary committee decided to perform chemoembolization and a liver resection, confirming HCC with cirrhosis in the surgical specimen. Two years later, the HCC relapsed and the committee decided to perform a liver transplantation with prior chemoembolization. The histological examination of the explant identified GSD, with cirrhotic nodules and HCC (Fig. 1). The patient had a good evolution 18 months after the transplant, with normal liver function and stability of the muscle and cardiac involvement.

Discussion

GSD-III patients would benefit from analytical and imaging follow-up in order to detect early liver complications and therefore, may have more therapeutic options. Liver transplantation is indicated in GSD-III with decompensated cirrhosis or HCC not amenable to local or surgical treatment. This is a good therapeutic option according to current experience. There are 6 published cases of liver transplantation in adults with GSD-III and cirrhosis (1-3) (Table 1), three had HCC in the explant. We present the first liver transplantation in an adult with GSD-III and cirrhosis in Spain.

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Table 1. Demographic and clinical characteristics of adult patients (> 15 years) with type III glycogen storage disease undergoing liver transplantation reported to date

	Gender	*Age*	*Glycogen storage disease type*	*Transplant indication*	*Presence of hepatocellular carcinoma*
Haagsma et al. Hepatology 1997	Female	33	IIIb	Decompensated cirrhosis	Hepatocellular carcinoma (2.5 x 3 cm) in the explant
Kondo et al. Clin Chim Acta 2013	Male	21	IIIa	Cirrhosis	No
Sentner et al. J Inherit Metab Dis 2016	1 male 3 female	15-35	3 IIIa 1 IIIb	Cirrhosis	Two patients had hepatocellular carcinoma in the explant
Iglesias et al.	Male	51	IIIa	Compensated cirrhosis and hepatocellular carcinoma	Hepatocellular carcinoma (2 cm)

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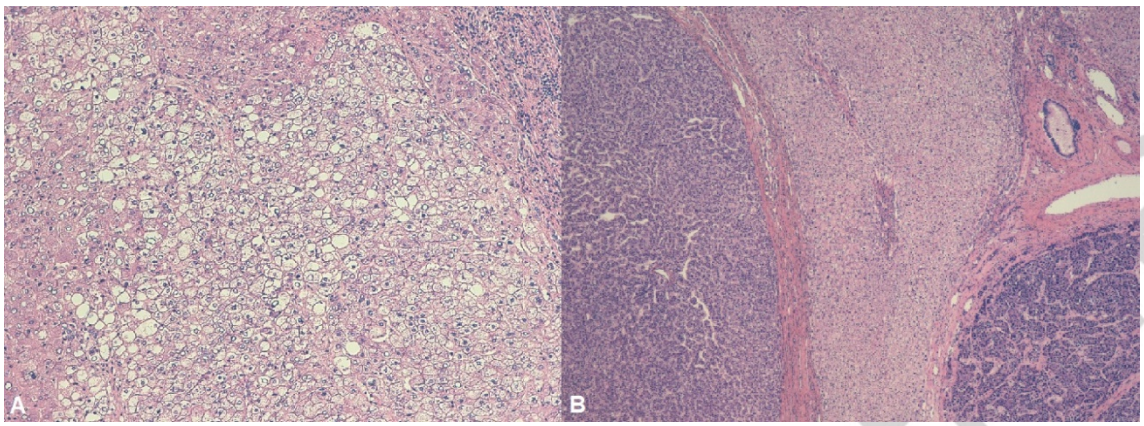

Fig. 1. Histology of the liver explant. A. Cells with a clear, swollen cytoplasm and vacuolated / clarified nuclei are observed (hematoxylin-eosin stain, 10x). B. Moderately differentiated hepatocellular carcinoma, with a central area of hepatic parenchyma that is unstructured and compatible with cirrhosis. Cells with a clear cytoplasm cells are also observed (hematoxylin-eosin stain, 4x). Two subcentrimetic satellite nodules of hepatocellular carcinoma were found, without microvascular tumor invasion.