HEPATOCELLULAR CARCINOMA (HCC) OCCURRENCE IN A LYSOSOMAL ACID LIPASE DEFICIENCY (LALD)-AFFECTED 49-YEARS OLD NON-CIRRHOTIC MALE: HOW TO MANAGE THE MALIGNANCY IN THIS RARE LIVER-DISEASE CONTEXT?

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A case report describes hepatocellular carcinoma development in a 49-year-old male with lysosomal acid lipase deficiency before cirrhosis onset, a previously unreported disease stage for HCC in this rare metabolic disorder.

KEY POINTS

- Lysosomal acid lipase deficiency is a rare autosomal-transmitted metabolic disease causing uncontrolled accumulation of cholesterol esters and triglycerides, leading to multi-organ damage and premature death.
- A 49-year-old male with statin-unresponsive dyslipidemia, juvenile myocardial infarction at age 27, chronic diarrhea, and hepatomegaly was diagnosed with a 30 mm hepatocellular carcinoma confirmed by biopsy.
- Dried Blood Spot testing showed low LAL activities and genetic testing confirmed LALD diagnosis; esophagogastroduodenoscopy revealed duodenal mucosa with marked foam cell infiltrate.
- The patient underwent microwave thermoablation as liver transplantation was not indicated due to hepatic disorder recurrence, and died after a subsequent myocardial infarction before receiving enzyme replacement therapy.
- This case suggests HCC can occur in non-cirrhotic LALD contexts, expanding the recognized spectrum of hepatic complications in this rare disease.

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