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Liver INTERNATIONAL WILEY

LETTER TO THE EDITOR

The time has come to look for metabolic dysfunction-associated fatty liver disease in adult patients with type 1 Gaucher disease

Dear Editors,

We thank Starosta and colleagues for allowing us to clarify some peculiar aspects of metabolic dysfunction-associated fatty liver disease (MAFLD) in type 1 Gaucher disease (GD1).[1,2]

MAFLD encompasses a broad spectrum of histological lesions spanning steatosis, ballooning and inflammation with/without fibrosis. We agree that steatosis with necro-inflammation, featuring steatohepatitis, more likely progresses to clinically significant liver fibrosis than histological 'simple' steatosis. However, according to current guidelines, the screening approach for MAFLD in at-risk populations and the diagnostic work-up in patients with suspected MAFLD should start with non-invasive evaluation of steatosis, followed by non-invasive assessment of fibrosis. While steatosis and fibrosis could be evaluated by non-invasive techniques, steatohepatitis diagnosis still exclusively relies on liver biopsy, that is unsuitable for screening purposes and is reserved for cases with suspicion of significant disease.[3,4]

The primary objective of our study was to verify if GD1 adults could be a population at risk of MAFLD by evaluating prevalence of and risk factors associated with steatosis. By showing that 40% of GD1 adults have non-invasive evidence of liver steatosis that is associated with features of metabolic syndrome (and not with GD1-related variables), we think that our results do demonstrate that stable GD1 patients are a population worthy to be screened for the presence of MAFLD.[1]

The assessment of liver disease severity in such a population is another aspect that deserves attention. In our studies we showed that liver fibrosis is common in GD1 adults (19%) and is significantly associated with metabolic risk factors and steatosis.[1,5] Consequently, we speculated that MAFLD may be an overlooked contributor of significant liver disease in stable GD1 patients. We recognize that the peculiar metabolic abnormalities associated with GD1 itself, and only partially reverted by GD1 treatment, may participate in predisposing GD1 patients to liver disease progression. Moreover, we agree that only liver biopsy is able to definitely stage liver disease severity and to determine if steatohepatitis is the underlying histological feature of liver disease progression in these patients. Further studies are eagerly awaited to resolve these issues. In the meanwhile, far from wanting to oversimplify the complexity of liver disease in GD1 and to prove causality, we think that the main messages from our studies are that GD1 patients, even if properly treated for their inherited disease, should be closely monitored for the development of metabolic risk factors, liver steatosis and fibrosis, and actively educated to healthy lifestyles.

CONFLICT OF INTEREST

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