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Hepatopathy of Mauriac syndrome. The importance of therapeutic adherence

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Ethical considerations: the authors declare that they have followed the protocols established by their respective health centers to access the data from the medical records in order to be able to carry out this type of publication for the purpose of research/dissemination for the scientific community. However, this article does not contain patient data.

Keywords: Mauriac syndrome. Hepatopathy. Diabetes.

Dear Editor,

We report the case of a 34-year-old female with a personal history of type 1 diabetes mellitus with very poor metabolic control and multiple microvascular complications, nephropathy, retinopathy, and polyneuropathy. She presented to the Emergency Room due to a headache and nausea, observing alterations compatible with diabetic ketoacidosis in the laboratory tests. Moreover, the tests revealed a HbA1c of 15 %, which was the reason why she was admitted to the ward. During admission, treatment with intravenous insulin was started and later with a bolus-basal regimen, with improvement in glycemic control.

Nine days after admission, she presented symptoms of edema in all four extremities,

distension and abdominal pain. On physical examination, she presented a height smaller than expected (150 cm), slight overweight (59 kg) and extensive hepatomegaly. Liver function tests showed an increase in transaminases (aspartate aminotransferase [GOT] 359 IU/l, alanine aminotransferase [GPT] 210 IU/l) and alkaline phosphatase (190 IU/l). Viral and autoimmune etiology was ruled out. An abdominal ultrasound was performed, showing a large hyperechogenic homogeneous hepatomegaly, so a percutaneous liver biopsy was performed (Fig. 1). This revealed liver parenchyma with preserved architecture, and hepatocytes with a large, clear cytoplasm, with a striking membrane and some empty nuclei with a glucogenic appearance.

Six months later, she went to the clinic and showed clinical and analytical improvement, with normalization of liver function and an abdominal ultrasound showing resolution of the hepatomegaly. She also presented good metabolic control, with glycated hemoglobin of 6.6 % and a time in range of 73 % without hypoglycemia.

Discussion

The Pierre Mauriac syndrome described in 1930 is characterized by growth failure, cushingoid appearance, hepatomegaly and hypertransaminasemia, in a patient with chronic uncontrolled DM1. The most common age of presentation is adolescence, although cases have been described in both children and adults. The hallmark of this syndrome is extreme liver enlargement from massive accumulation of glycogen. The diagnosis of hepatopathy requires high clinical suspicion and the presence of glycogen accumulation must be corroborated with a liver biopsy. The accumulation of glycogen in hepatocytes is partly caused by long periods of hyperglycemia, in which glucose enters the hepatocyte independently of insulin and is converted to glycogen (1-4).

Mauriac syndrome is currently a rare cause of liver disease, due to improvements in the control and treatment of patients with DM1. However, some cases are described in individuals with complicated social situations or without therapeutic compliance. This is a reversible condition after improvement in glycemic control with adequate insulin control (1-3).

For this reason, we believe it is important to suspect this clinical scenario in patients

with poor glycemic control and symptoms of pain and abdominal distension.

References

1. Fitzpatrick E, Cotoi C, Quaglia A, et al. Hepatopathy of Mauriac syndrome: a retrospective review from a tertiary liver centre. Arch Dis Child 2014;99(4):354-7. DOI: 10.1136/archdischild-2013-304426

2. Kim MS, Quintos JB. Mauriac syndrome: growth failure and type 1 diabetes mellitus. Pediatr Endocrinol Rev 2008;5(Suppl 4):989-93.

3. Elder CJ, Natarajan A. Mauriac syndrome: a modern reality. J Pediatr Endocrinol Metab 2010;23(3):311-3. DOI: 10.1515/JPEM.2010.23.3.311

4. MacDonald MJ, Hasan NM, Ansari IU, et al. Discovery of a genetic metabolic cause for Mauriac syndrome in type 1 diabetes. Diabetes 2016;65(7):2051-9. DOI: 10.2337/db16-0099

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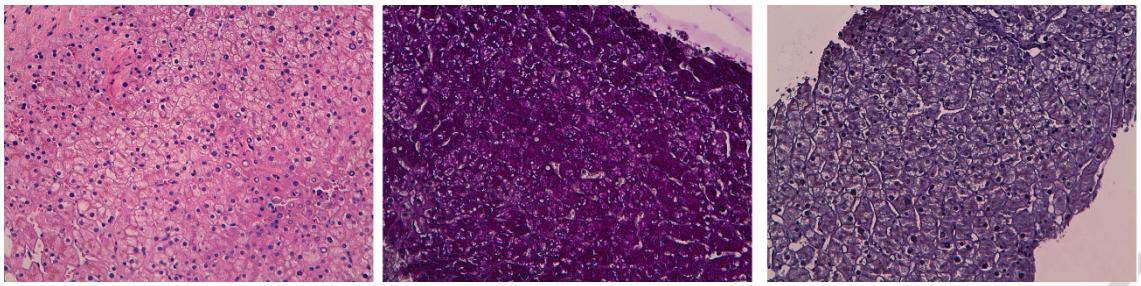

Fig. 1. A. 20x hematoxylin-eosin staining, showing hepatocytes with a clear cytoplasm and striking membranes, and some clear nuclei with a glucogenic appearance. B. PAS staining that demonstrates the accumulation of glycogen in the cytoplasm staining intense pink. C. Clear cytoplasm in the PAS diastase staining, demonstrating that it is glycogen.