More about Renal Disease in Type Ia Glycogen Storage Disease

S.T.M.	Tomé Martínez	de Rituerto ^a
D.		Sanchez-Guisande ^b
J.		Forteza ^c
D.		Novoa ^b
R.		Romero ^b
M.I.		Rodríguez ^a
E.		Barrio ^a

Servicios de ^aMedicina Interna, ^bNefrología, ^cAnatomía Patológica, Hospital General de Galicia, Santiago de Compostela (Spain)

Dr. Santiago Tomé Martínez de Rituerto, Avenida Rodríguez de Viguri, 45-3° B, E-15704 Santiago de Compostela (Spain)

Dear Sir,

Type Ia glycogen storage disease (GSD) is characterized by a deficiency in glucose-6-phosphatase, with the resulting accumulation of glycogen in the liver, kidney and intestine [4,5]. The development towards renal insufficiency was rarely documented, and only recently some cases with a fatal evolution have been described.

We are presenting a new case in which histological changes and their relationship with metabolic alterations are analyzed. It is the case of a 30-year-old man sent to us because he showed hyperlipemia. In his childhood, hepatopathy due to liver fat degeneration was diagnosed. He followed an irregular dietetic treatment, showing a delayed growth as only lateration.

When he presently was entered for ultra-sonographic examination of his abdomen, he showed a great hepatomegaly with a hete-roechogenic image inside. The right and left kidney measured 13 and 14 cm, respectively.

Total protein, BUN, creatinine and uric acid were normal. He had glycemia, 55 mg/dl; triglycerides, 295 mg/dl; cholesterol, 585 mg/dl, and lactic acid, 34 mg/dl (normal 5-15 mg/dl). After glucagon stimulation, the glycemic level rose to 59 and lactic acid to 84 mg/dl. Protein in urine in 24 h was 5 g and creatinine clearance 120 ml/min.

Renal biopsy showed undetectable levels of glucose-6-phosphatase. The endothelial capillary as well as the tubular cells showed introcytoplasmic vacuoles that are proved to be ultrastructurally glycogen (fig. 1). The

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Fig. 1. Ultramicrograph of glycogen deposits in renal tubules. ×6000.

characteristic finding in renal histology is focal and segmentary sclerosis. Proteinuria in the early phases indicates that the glomerular comes before the interstitial damage, whereas a high clearance would speak in favor of hyperfiltration playing a certain role [3]. In this sense, nowadays it is known that the increase in contraregulating hormones sec-

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ondary to chronic hypoglycemia is the cause of hyperfiltration. Reviewing the different series proves that it is precisely those patients following an irregular treatment who develop stronger renal alterations, as in the case we are dealing with [1, 6].

The therapeutic advance represented by liver transplantation in these patients, reversing renal alterations, among others, has been recently described [7, 8]. In any case, the bases of the treatment are a careful dietetic treatment, a strict metabolic control and a strict follow-up of the renal function.

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