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CORRESPONDENCE

Pseudohypertriglyceridemia: A Diagnostic Conundrum

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To the Editor: Hyperglycerolemia due to glycerol kinase deficiency (GKD) may be misdiagnosed as hypertriglyceridemia and treated unnecessarily with lipid-lowering drugs.

A 13-y-old boy was incidentally detected to have elevated triglycerides (TG, 518 mg/dL) and treated aggressively with lipid-lowering drugs (Rosuvastatin, fenofibrate, and ezetimibe) over the preceding year without much response. There was no prior history of any systemic illness, drug intake, or family history of dyslipidemia. Physical examination, including fundus examination, was unremarkable. There were no xanthomas. Visual inspection of blood samples showed clear serum. The photometric lipemic assay (OSR62166, Beckman Coulter) was negative. Serum TG concentrations were elevated (433 mg/dL), but total cholesterol (135 mg/dL), high-density lipoprotein cholesterol (44 mg/dL), low-density lipoprotein cholesterol (103 mg/dL), amylase and lipase were normal. Secondary causes, including thyroid, hepatic, and renal disorders were ruled out. Glycerol blanking could not be performed. Clinical exome sequencing revealed a hemizygous start loss variant in exon 1 of the glycerol kinase gene (c.3G>A), affecting translation (p.Met1), confirming GKD. A lipid profile done four weeks after discontinuing medications showed no worsening of TG levels.

Pseudohypertriglyceridemia refers to overestimating TG levels by laboratory assays due to elevated plasma glycerol concentrations in metabolic disorders such as GKD [1, 2]. The clinical spectrum of GKD ranges from an infantile presentation with metabolic crisis, hypoglycemia, and developmental delay to an asymptomatic adult form, which is only incidentally diagnosed as hypertriglyceridemia [3]. The absence of visible lipemia in the serum sample, lack of consistent clinical manifestations, normal levels of other lipoproteins, and poor response to lipid-lowering agents should raise suspicion for pseudohypertriglyceridemia. Glycerol blanking can differentiate between artefactual and true TG elevation. Elevated serum (normal 5–20 mg/dL) and urine glycerol levels (normally undetectable) also support the diagnosis [4]. Recognition of this entity is important to avoid misdiagnosis, unwarranted treatment, and extensive testing.

Declarations

Conflict of Interest None.

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