TWO CASES OF "UNKNOWN" LIPOPROTEIN LIPASE (LPL) DEFICIENCY AND DIABETES MELLITUS

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Lipoprotein lipase (LPL) deficiency is an autosomal recessive disease with deficient extrahepatic removal of blood lipoproteins.

Primary LPL deficiency can be exacerbated by coexistent conditions such as diabetes, where relative or absolute insulin deficiency leads to an additional secondary LPL deficiency

We describe two cases in which primary LPL deficiency overlapped with previously diagnosed T2DM and with T1DM at onset, respectively.

17 year-old T2DM girl with positive family history for T2DM, obesity, mixed dyslipidemia and early cardiovascular diseases presented with central obesity (BMI 30.7 kg/m2, WC 99 cm), acanthosis nigricans, limb xanthomas.

HbA1c 123 mmol/mol

total cholesterol 562 mg/dl

triglycerides 2400 mg/dl

normal ApoA1 and ApoB

high levels of ApoB/A1 (1.94)

Abdominal US: enlarged, hepatic steatotis with focal nodular hyperplasia, an ovarian cyst.

7 year-old male with rapidly progressive polyuria, polydipsia, weight loss and impaired consciousness.

HbA1c 66 mmol/mol (C-peptide 0.12 ng/ml, beta-cell antibodies +) → T1DM

Triglycerides 11628 mg/dl → PA → 2510 mg/dl

Total cholesterol 1128 mg/dl → PA → 380 mg/dl

IV rehydration, insulin therapy, plasmapheresis
Non-ST segment elevation: a myocardial infarction was suspected.

Abdominal US: thickening of the gallbladder wall, with thick bile; hyperechogenic thinned pancreas.

3 heterozygous variants - Promoter - Exon 2 (Asp_9>Asn) - Exon 6 (Asn_{291}>Ser)	**LPL gene**	Heterozygous mutation – Exon 5 (Pro207>Leu)
No pathogenic mutation	**APOA5 gene**	Polymorphism – Exon 3 (Ser_{19}>Trp)
No pathogenic mutation	**APOC2 gene**	No pathogenic mutation

Disclosure statement: nothing to disclose