The role of continuous subcutaneous insulin infusion therapy in a case of Seip-Berardinelli congenital lipodystrophy

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Introduction

- Seip-Berardinelli congenital lipodystrophy (SBCL) is an extremely rare autosomal recessive disorder characterized by a congenital absence of adipose tissue.
- Hepatic steatosis, splenomegaly, skeletal muscle hypertrophy, hypertrophic cardiomyopathy, insulin-resistance and diabetes mellitus (DM) are some of the features of such patients.

Case report

- A caucasian-male patient, currently with 21 years-old, was referred at 6-months-age because of “muscular phenotype” and liver function alterations. On examination, muscular appearance with minimal subcutaneous fat (Figure 1) and hepatosplenomegaly were identified. Laboratorial tests revealed abnormalities in cholestatic liver enzymes; glycemia was normal. Ultrasound exams documented diffuse liver steatosis and hypertrophic cardiomyopathy. The identification of heterozygous BSCL2 gene mutation (p.Pro65ArgfsX28 and p.Thr109AsnfsX5) established the diagnosis of SBCL type 2.
- At 7 years old it was diagnosed DM (fasting plasma glucose= 182mg/dL; HbA1C=8.8%; insulin=116.7mU/L; C-peptide=8.7 ng/mL; HOMA-IR=52.4). He was initially treated with metformin and later at 11 years old with concomitant insulin.
- He had always a poor metabolic control (Figure 2) justifying, at 16-years old age the start on continuous subcutaneous insulin infusion (CSI) therapy and a significant improvement was obtained (last HbA1C=8.0%). He is currently under metformin (4 g/day) and CSI therapy (daily dose of insulin of 155U).

Discussion

- We report a rare case of lipoatrophic DM in childhood associated with severe insulin-resistance within the context of SBCL. In this setting, DM is frequently difficult to control and the management may involve insulin-sensitizers and exogenous insulin.
- Although the CSI therapy is normally used in insulin-deficient patients, classically type 1 DM, some insulin-resistant DM patients, as the one reported, may benefit from this therapeutical approach.