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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* (CSII) therapy and a significant improvement was obtained (last HbA1C=8.0%). He is currently under metformin (4 g/day) and CSII therapy (daily dose of insulin of 155U).

Figure 1: Old picture of the patient with the age of 11 months

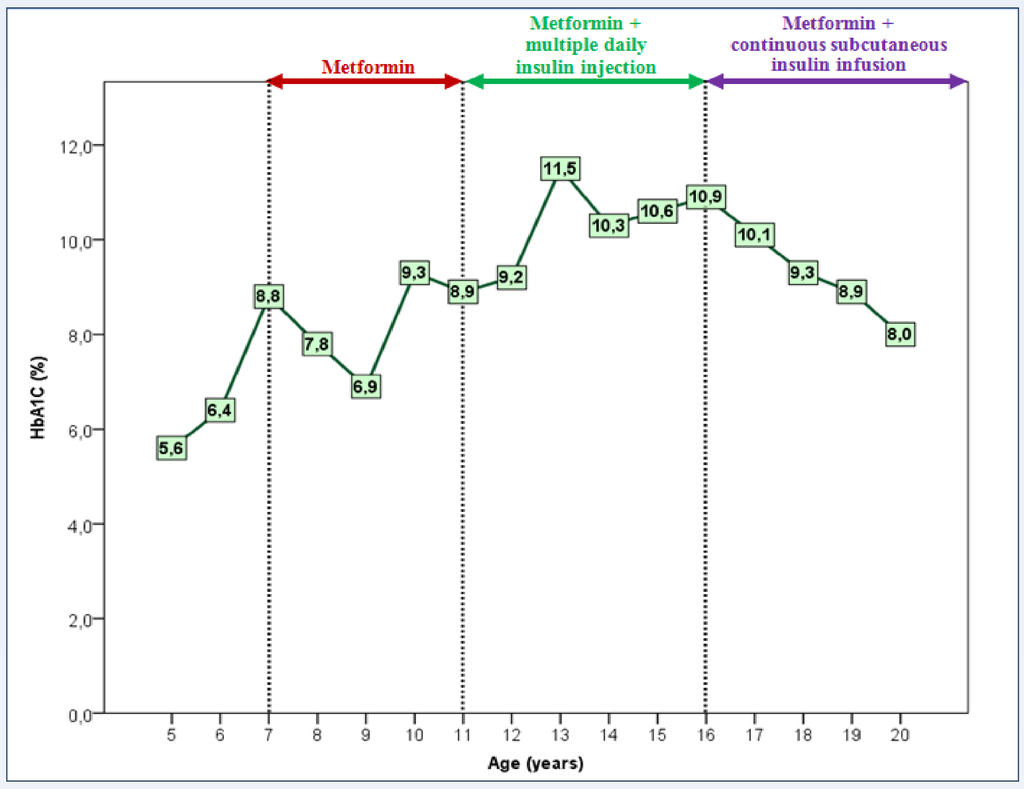

Figure 2: Evolution of HbA1C levels during the disease course
Therapeutic approaches are indicated at the top of the figure.

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 CSII 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.