BERARDINELLI SEIP CONGENITAL LIPODYSTROPHY. A LIGHT OF HOPE.

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The lipodystrophy syndromes are a heterogeneous group of congenital or acquired disorders characterized by either complete or partial lack of adipose tissue (lipoatrophy). Berardinelli Seip congenital lipodystrophy (BSCL) is a rare metabolic disorder characterized by severe generalized lipodystrophy since birth, insulin resistance and dyslipidemia since early infancy.

We report a 6 year old girl who arrived from Pakistan with the diagnosis of Diabetes Mellitus. Almost from birth she presented generalized absence of fat and a prominent abdomen. Since the year before polyuria and polydipsia appeared, starting with subcutaneous insulin.

Clinical features: absent of adipose tissue almost completely except on mouth, palms, soles and scalp. Protuberant abdomen due to 7 cm hepatomegaly. Acanthosis nigricans was present.

Biochemical analyses: glucose 14.8 mmol/L, normal total cholesterol concentration, triglyceride 4.96 mmol/L, HbA1c 12%, insulin 5.3 μU/mL, leptin 1.7 ng/mL.
Gen mutation: AGPAT2 c.755_763delTGAGGACCA.
After a year without treatment serum triglyceride raised to 6.45 mmol/L, glucose 16.6 mmol/L and HbA1c 13.6%.

Conclusion:
Human recombinant leptin is effective for controlling diabetes, hypertriglyceridemia and hepatic steatosis. Positive effects are notorious since the beginning of the treatment. No remarkable adverse effects where observed.