54th Annual Meeting 1-3 October
ESPE 2015 BARCELONA
European Society for Paediatric Endocrinology

BERARDINELLI SEIP CONGENITAL LIPODYSTROPHY. A LIGTH OF HOPE.

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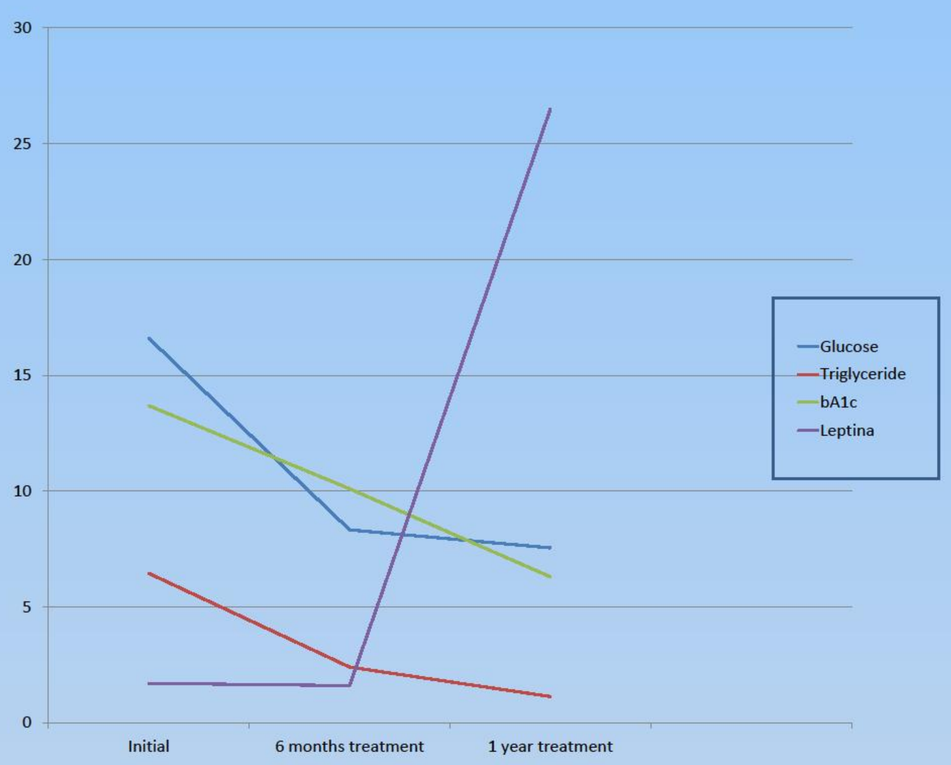
The lipodystrophy syndromes are a heterogeneous group of congenital or acquired disorders characterized by either complete or partial lack of adipose tissue (lipoatrophy).Berardinelli Seip congenital lipodystrophy (BSCL) is a rare metabolic disorder characterized by severe generalized lipodystrophy since birth, insulin resistance and dyslipemia 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: glucose14,8mmol/L, normal total cholesterol concentration , triglyceride 4,96mmol/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%.



Before

After

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.

ESPE 2015
355--P2
Fat Metabolism, Obesity
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DOI: 10.3252/pso.eu.54espe.2015
Poster presented at: 54th Annual Meeting 1-3 October ESPE 2015 BARCELONA European Society for Paediatric Endocrinology