SEIP-BERARDINELLI SYNDROME

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BACKGROUND

Seip-Berardinelli syndrome is a rare form of congenital lipodystrophy.

OBJECTIVE AND HYPOTHESIS

To report a patient later diagnosed with Seip-Berardinelli syndrome referred initially for evaluation due to low weight gain.

POPULATION AND/OR METHODS

We performed the report of the case along with a literature review.

RESULTS

On physical examination, at 3 years and 8 months, her weight was 19 kg (P90-97); length was 108 cm (P90-97) and head circumference was 49 cm (P50).

She had general reduction of subcutaneous tissue, leaving the musculature visible; acanthosis nigricans in neck and axillary regions; umbilical hernia and hirsutism in face, back and limbs (Figures 1 and 2).

Her laboratory tests showed VLDL dosage of 166 mg/dL (Normal up to 40), triglycerides of 829 mg/dL (Normal up to 160), cholesterol of 151 mg/dL (Normal up to 200); glucose of 83 mg/dL (Normal: 70 to 110) and insulin of 37 IU/mL (Normal <30).

Echocardiography showed concentric hypertrophy of the left ventricle.

CONCLUSIONS

Clinical and laboratory findings were compatible with the diagnosis of Seip-Berardinelli syndrome.

These patients have almost complete absence of adipose tissue from birth, which causes them to have an undernourished appearance.

The treatment of this disease is based on diet and drug therapy for dyslipidemia control and avoiding its consequences.