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

The patient was referred due to low weight gain. She was the second daughter of a non-consanguineous couple.

She was born vaginally at 36 weeks of gestation, measuring 46 cm, weighing 2685 g, with head circumference of 32 cm and Apgar scores of 9 and 10 at first and fifth minutes.

The patient developed neuropsychomotor and speech delay.

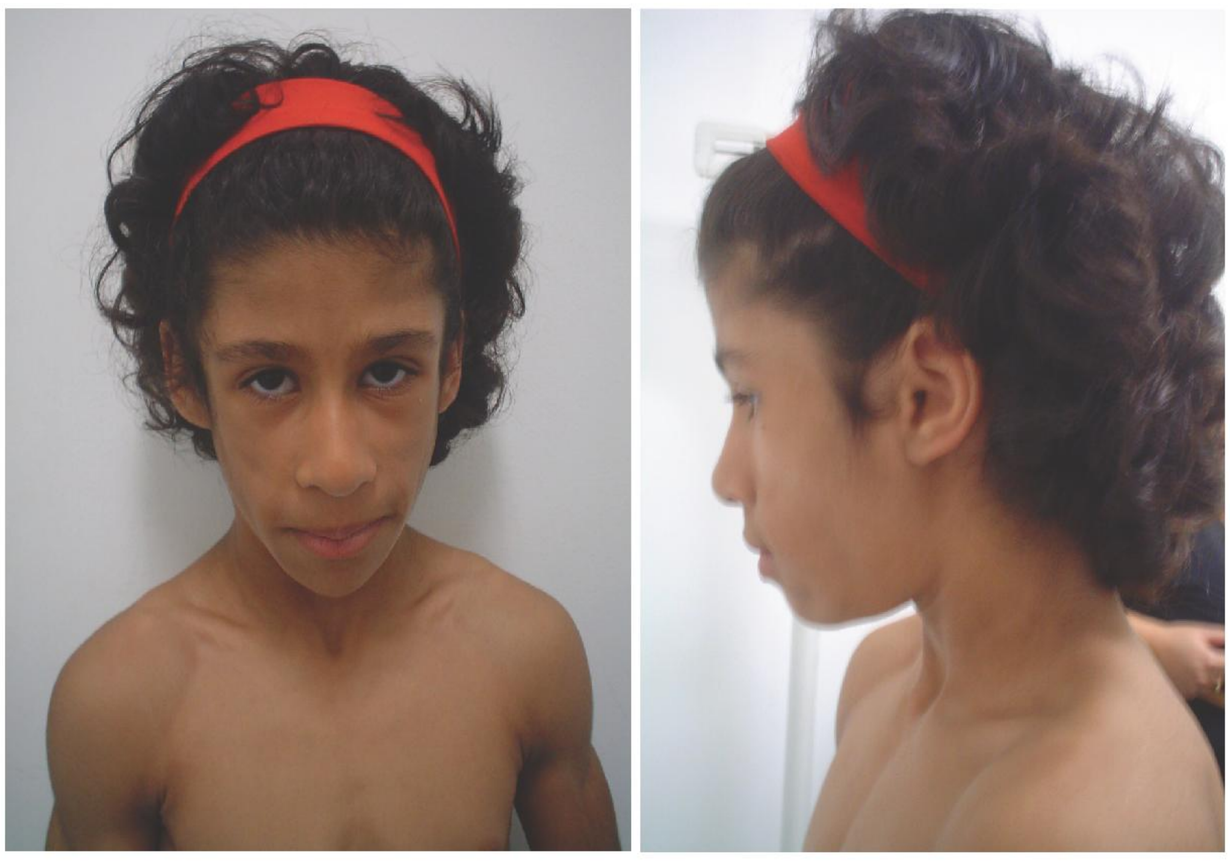

Figure 1. Craniofacial appearance of the patient at 5 years of age.

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.

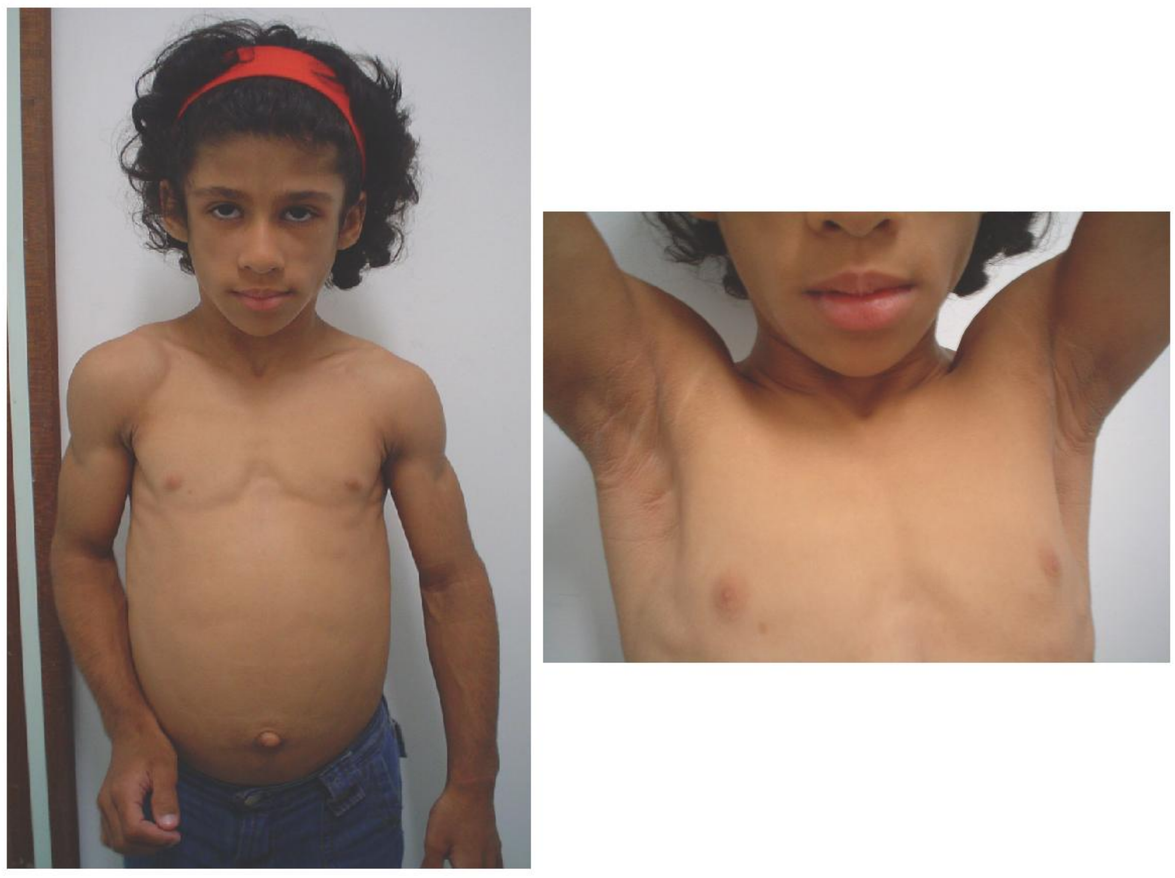

Figure 2. Note general decrease in subcutaneous tissue, leaving the musculature most visible, as well as acanthosis nigricans in axilla.

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.

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Diabetes 1

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