SLEP SUMMER SCHOOL 2020

Fellow 5. Aline Dantas Costa Riquetto

A 2-years-old (y) girl presented with low weight gain and hypertriglyceridemia since 1y. She was adopted at 2 months old. The adoptive mother referred that even with adequate food intake there was no satisfactory weight gain (aspect of malnutrition). She also referred accumulation of muscle mass mainly in limbs. The mother had no data on birth parents. Birth weight was 2.500 Kg, birth length 43.5 cm and head circumference 33 cm at 37 weeks of gestational age.

The clinical exam revealed acromegaloid facies, enlarged hands and feet, generalized lack of adipose tissue (but preserved in the palms and soles), muscular hypertrophy and prominent veins. She didn't have acanthosis *nigricans*. Tanner pubertal stage: M2P2. Weight and height was appropriate for age. Systemic examination was normal.

Laboratory tests showed: total cholesterol 4.5 nmol/L (173 mg%); LDL 2.2 nmol/L (83 mg%); HDL 0. 5 nmol/L (19.3 mg%); triglycerides 3.9 nmol/L (345 mg%); insulin 9 pmol/L (1.3 mIU/L); fasting plasma glucose 4 mmol/L (72 mg%); HgbA1c 29.0 mmol/mol (4.8%); LH < 0.1 IU/L; FSH 3,1 IU/L; androstenedione 4.9 nmol/L (1.4 ng/ml); estradiol: undetectable; total testosterone < 0.4 nmol/L (<10 nf/dl); leptin 0.9 µg/L.

Transabdominal ultrasound revealed homogeneous liver echotexture with liver size at the upper normal limit. Echocardiogram was normal.

A targeted next-generation sequencing allowed to confirm the presumptive diagnosis.


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