AN UNUSUAL CAUSE OF NEONATAL HYPERGLYCEMIA – CASE REPORT

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INTRODUCTION

Hyperglycemia is a common event in neonates, frequently associated with specific clinical conditions (sepsis, drugs or intravenous fluids) other than neonatal diabetes. Unusual endocrinometabolic syndromes must be considered whenever initial studies are inconclusive.

CASE REPORT

NEWBORN NICU ADMISSION

- Low birth weight
- Hypotonia
- Feeding dificulties

PRENATAL HISTORY

Healthy parents
IG IP 37WG Late IUGR
Oligoamnios / Fluxometric changes
Maternal hypertension (35 W) and autoimmune thyroiditis

BIRTH

Eutocic delivery Apgar 8/10
W: 1710 g (<<10) H: 43 cm (<<P10)
HC: 29 cm (<<10)

DAY11

Tachycardia
Hyperglycemia
180 - 349 mg/dL

CBC, venous blood gas
Blood cultures
Ketonemia/Ketonuria

Negative

Enteric pause
IV fluids (glucose 5mg/Kg/min)
Insuline therapy for 24 hours

DAY 28

Tachycardia
Progressive hypotonia

Hyperglycemia
200 mg/dL

Hypertrygliceridemia
3549mg/dL

Enteric pause (< 24h)
Very low lipid formula
Diet supplementation with soy and corn oils (gradually added)

DAY 34

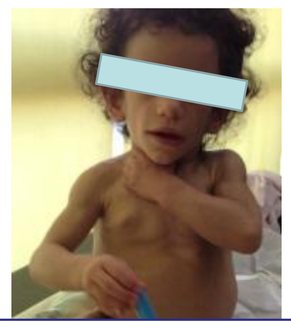
Discharged home
TG levels - 175mg/dL
Net weight increase 20g/day

Exclusion of main causes of primary hypertrygliceridemia:

- Apo A1, B48, CI and CII: normal
- LPL, APOC-II, APOAV, GPIHBP1 and LMF1 genes: no pathogenic mutations

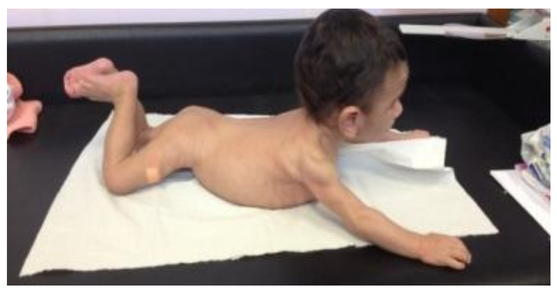
FOLLOW UP

- Lipodystrophic phenotype gradually developed
- Insulin levels persistently elevated (no hyperglycemia)
- Hepatomegaly and steatohepatitis
- TG 400-800mg/dl



BSCL2 gene sequenced: Compound Heterozygous (c.399C>A (p.Tyr133*) and c.604C>T (p.Arg202*)

BERARDINELLI-SEIP SYNDROME



At 15 months
Borderline inferior psychomotor development
Breatst button without precocious puberty and neck *acanthosis nigricans*
Normal growth velocity, and concordant skeletal age
Concentric hypertrophic cardiomyopathy (IV septum 9mm)
Under diet with 11% lipid, 15% protein and 74% carbohydrates + lcPUFAs

DISCUSSION

The authors present a case of Berardinelli – Seip Syndrome (Congenital generalized lipodystrophy type 2) as the primary cause of neonatal insulin resistance and hyperglycemia. This syndrome is characterized by the absence of functional adipocytes with storage of lipids in muscle and liver and consequent hepatomegaly, steatosis, and skeletal muscle hypertrophy. Besides early insulin resistance, other endocrine manifestations that should be regularly surveilled include diabetes mellitus of difficult control, hirsutism, and precocious puberty.

REFERENCES

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