UNEXPECTED CLINICAL FEATURES IN A FEMALE PATIENT WITH PROPIOMELANOCORTIN (POMC) DEFICIENCY

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The authors declare there is no conflict of interest

Background: Proopiomelanocortin (POMC) deficiency is characterized by severe, early-onset hyperphagic obesity and congenital adrenal insufficiency, the latter secondary to corticotrophin (ACTH) deficiency. We presented a patient with POMC deficiency and neuromotor retardation coexistance.

Case: 3,5 years old girl patient presented to the clinic with complaint of
- Rapid weight gain
- Hypoglycemic seizures (Figure 1 and 2)

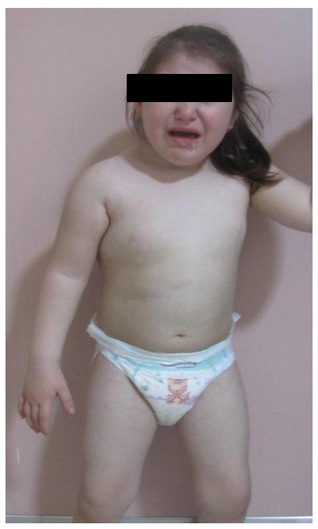

Figure 1

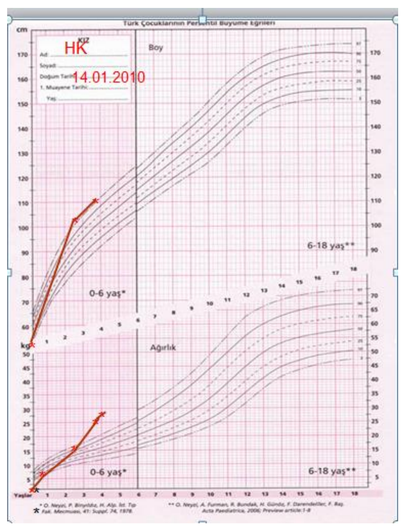

Figure 2

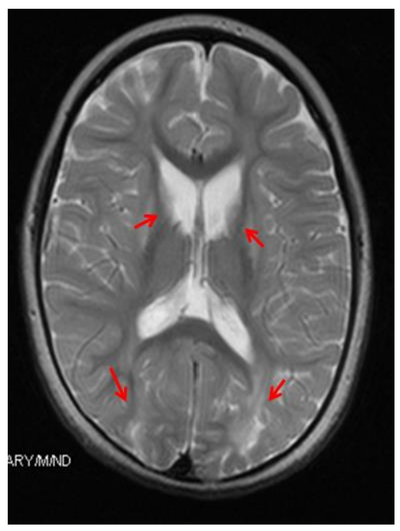

Figure 3

History:
- Birth weight 3100 g
- First offspring of consanguineous parents
- Neonatal cholestasis and hypoglycemic seizures (serum glucose: 30 mg/dl)
- After five months; rapid weight gain and decline in neuromotor development stages
- Treated in Pediatric Intensive Care Unit twice (septic shock due to pneumonia, severe hypoglycemia)

History:

<u>Neurometabolic disease was investigated because of;</u>
- Motor mental retardation, ataxia, bilateral hyperintense lesions in the basal ganglia in cranial magnetic resonance imaging (Figure 3) and increased lactate-lipid peak in proton magnetic resonance spectroscopy

- Serum electrolyte, glucose, insulin, lipids
- Blood aminoacids, carnitine profile,
- Lactate and ammonia levels
- Urinary organic acids analysis

→ **NORMAL**

Endocrinology consultation ;

Rapid weight gain

Obesity

Episodes of hypoglycemia at the time of infection

Physical exam:
- Weight: 26 kg (SDS: 3.02)
- Height: 110 cm (SDS: 3.07)
- Body mass index: 21.5 kg/m^2 (SDS: 3.12)

- Red hair, pale skin, ataxic gait

Laboratory:

ACTH (pg/ml)	**<10**
Kortizol (µg/dl)	**0.01**
17 OH progesteron (ng/ml)	**<0.01**
sT4(ng/dl)	**0.76**
TSH (µIU/ml)	**6.89**
PRL (ng/ml)	**22,26**

Diurnal rhythm	**08:00 am**	**23:00 pm**
ACTH (pg/ml)	**<10**	**10.6**
Cortisol (µg/dl)	**<0.4**	**<0.4**

	baseline	**5. mn**	**15. mn**	**30.mn**	**60. mn**
ACTH (pg/ml)	**13**	**11**	**15**	**14**	**14**
Cortisol (µg/dl)	**0.06**	**0.07**	**0.01**	**0.01**	**0.07**

CRH stimulation test

- **Molecular analysis of POMC gene** : a homozygous mutation, stop kodon (c.64delA/pMet22TrpfsX49)

- Her parents were heterozygote

- **Conclusion:** Severe motor mental retardation and cranial MRI pathology in patients with POMC deficiency haven't been reported previously in the literature. Bilateral hyperintense lesions in the basal ganglia and the increased lactate-lipid peak was thought to be the result of recurrent hypoglycemia.