A CASE WITH CENTRAL ADRENAL INSUFFICIENCY AND EARLY ONSET OBESITY: PROOPIOMELANOCORTIN DEFICIENCY

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Introduction

- Proopiomelanocortin (POMC) deficiency is a rare disease characterized by central adrenal insufficiency, early onset obesity, red hair, and impaired skin pigmentation. This disease is caused by mutations of *POMC* that is localized in 2p23.3.
- Here, we aimed to present a case with central adrenal insufficiency, red hair, and rapid weight gain, and who was detected frame shift mutation in the *POMC*.

A 16-day-old girl

- History: She was brought to our hospital due to poor feeding and jaundice. Her past medical history revealed that she was born after the first and uneventful pregnancy of mother with a birth weight of 3270 grams at 41 gestation weeks. Family history was unremarkable and the parents were no relatives.
- Physical Examination: Her weight was 3030 g (-1.29 SDS), height was 50.6 cm (-0.17 SDS), head circumference was 35.8 cm (0.14 SDS), and also icteric appearance and red hair was observed. Genital examination revealed normal female external genitalia.
- Laboratory: Hypoglycemia (30 mg / dL), mild hyponatremia, negative urine ketones; high serum levels of total bilirubin, ALT / AST, and elevated ammonia-lactate were found. Serum acyl / carnitine profile was in normal range. Hormonal profile revealed free T4 1.14 ng / dL, TSH 9.01 U / L, FSH 0.57 U / L (0.1-3.3), LH 0.24 U / L (0-1.9), DHEA-S 4.7 µg/dL, prolactin 18.5 mIU / L (3-24), serum insulin 0.32 U / L, cortisol 0.08 µg/dL, ACTH <5 pg / mL. The peak cortisol response was inadequate in the low-dose ACTH test (11.2 µg/dL).
- Imaging: Pituitary MRI was normal.
- Treatment: Central (secondary) adrenal insufficiency was established and 10 mg / m2 hydrocortisone was started.

Follow-up

- Hypoglycemia was not recorded after hydrocortisone treatment, and progressive weight gain was observed during the follow-up.
- At the most recent follow-up when she was at the age of three years and 10 months, weight was 23.2 kg (3.07 SDS), height was 101.2 cm (0.31 SDS) and body mass index was 22.7 kg / m2 (3.58 SDS).
- She was on treatment with hydrocortisone (6.8 mg/m^2/ day).

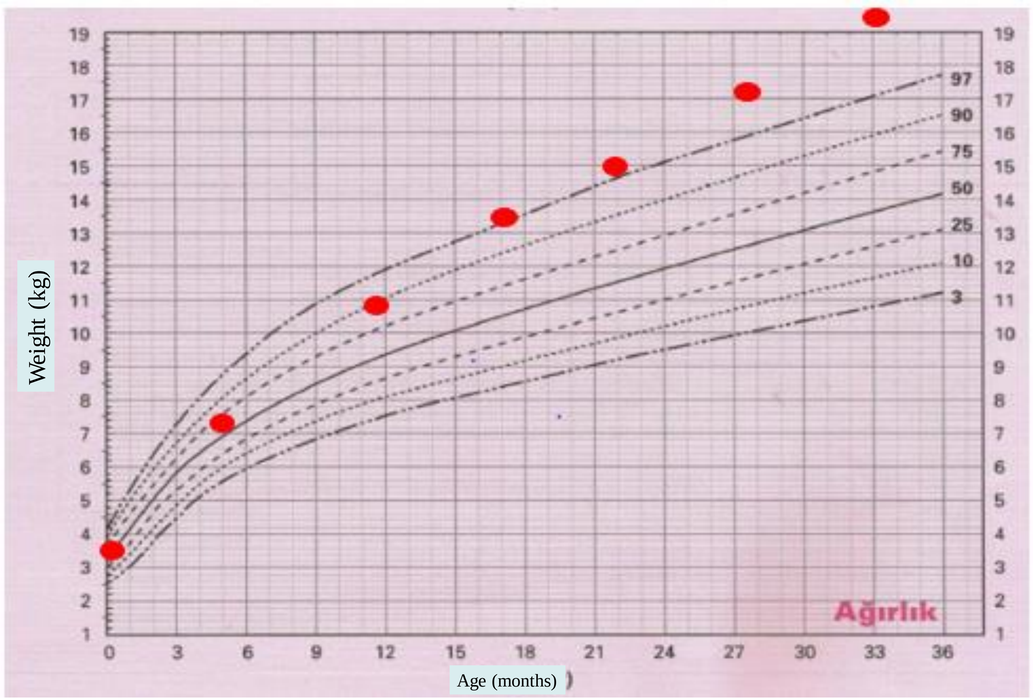

Figure: Demonstration of weight gain in the follow-up.

Genetic Analysis

- Central (secondary) adrenal insufficiency was established and 10 mg / m2 hydrocortisone treatment was started.
- Red hair and central adrenal insufficiency was suggestive for the diagnosis of POMC deficiency and following genetic analysis, homozygous mutation in the *POMC* [c.206delC (p.P69Lfs*2)] was detected.
- The parents were both heterozygous for the same mutation.

Conclusion

- In cases with central adrenal insufficiency, red-hair and early-onset obesity, POMC deficiency should be suspected and molecular genetic analysis of *POMC* should be performed..