

A new case of Proopiomelanocortin deficiency

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Non-disclosure statement

Introduction

The proopiomelanocortin is a polypeptide of many biologically active peptides involved in many key functions which have not yet been clarified. The mutation in the gene encoding this polypeptide is associated with a clinical trials characterized by early-onset obesity, secondary adrenal insufficiency and alteration of pigmentation. Eight cases with known genetic mutation have been published.

Case report

A 27 day old infant from North African:
hypoglycemia (from the fifth day of his life)

Physical examination

Red hair pigmentation + pale skin

Diagnostic studies

Genetics analysis

Novel homozygous mutation in the POMC* gene, in exon 3

Treatment

hydrocortisone + fludrocortisone.

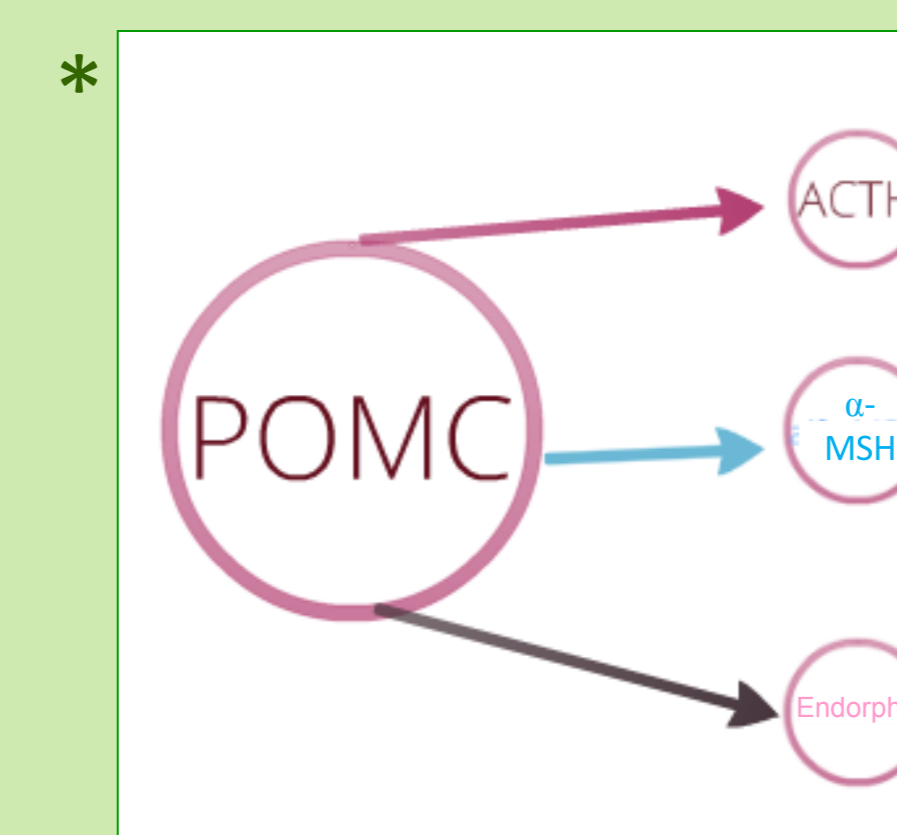
Evolution

A. The glyceamic profile was normalized after starting treatment.
B. He presented increased appetite with significant weight gain at 2 years (BMI +6.66DS and size: P94).

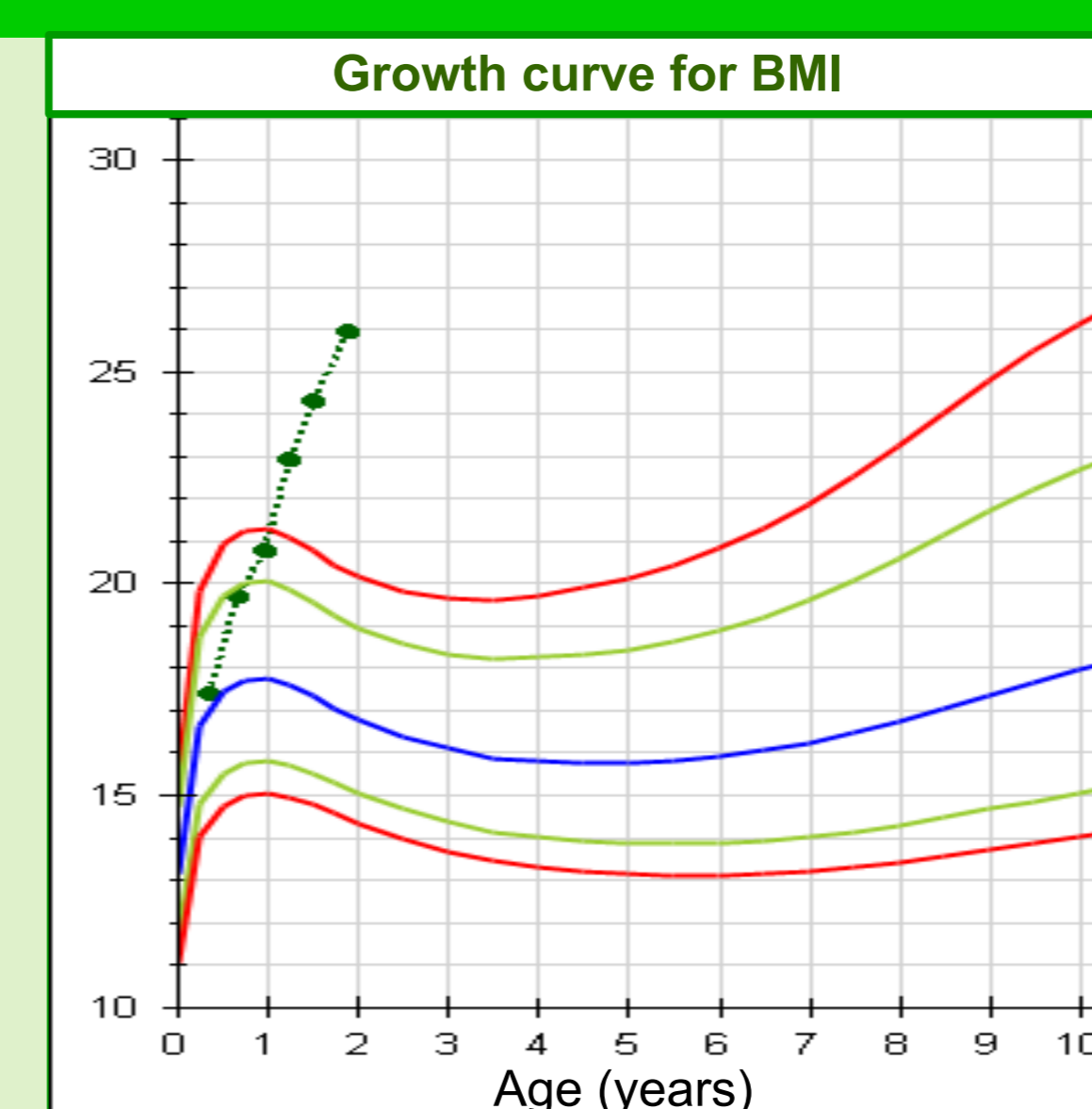
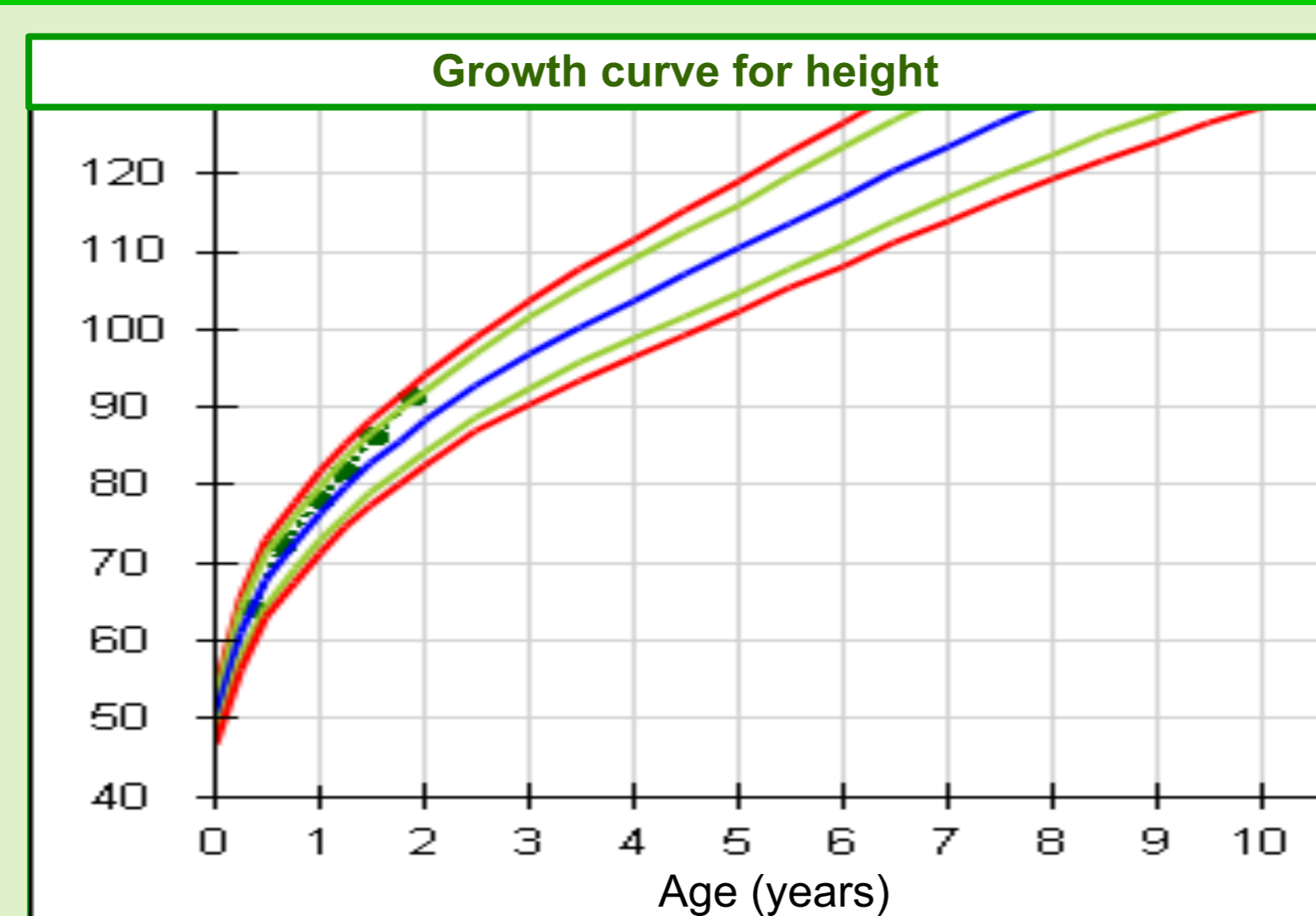
Laboratory tests

- TSH and free T4 and electrolytes: normal range
- ACTH < 5pg/ml
- Cortisol 0.4mcg/dl (N: 100-260)
- DHEAS < 1ng/ml (N: 28-852)
- Plasma Renin Activity 3.4ng/ml/h

Imaging studies: no pathologic features



Growth curves



Conclusions

1. We describe a new case of complete loss-of-function mutations of the POMC gene, manifested by the triad features of early obesity, hypocortisolism and pigmentation problems.
2. A new mutation of the POMC gene is described.
3. The analysis of the clinical characteristics of the patient can help the better understanding of the functions of these peptides, such as the leptin-melanocortin system, which could help in the understanding of obesity and possible therapeutic avenues.