A CASE OF EARLY-ONSET OBESITY WITH A NEW MUTATION AT THE PROOPIOMELANOCORTIN GENE

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INTRODUCTION

Proopiomelanocortin (POMC) is a 241-amino acid polypeptide encoded by the POMC gene located at 2p23.3. Complete mutation in the gene is associated with the classical triad of early-onset obesity, adrenal insufficiency, and red hair. A new POMC gene mutation was detected in our patient who presented with the hypoglycemia symptom in the second month of life. With this case, our information on the new mutation to phenotype relationship, leptin-melanocortin pathway, POMC-derived peptides and energy balance will be discussed.

CASE REPORT

The female patient aged 2.5 months was brought to another healthcare center with restlessness, cyanosis, cross eye and spasms. Her blood glucose was measured as 31 mg/dl and was therefore referred with the initial diagnosis of metabolic disease.

Born at 39 weeks with a birth weight of 3000 g, the patient had no problems during the prenatal/natal period. Her mother and father were not relatives.

At age 2.5 months

- Body weight: 3700 g (3p)
- Height: 51 cm (<3p)
- Head Circumference: 35 cm (<3p)
- Eyebrows and hair were red, External female appearance

Laboratory results:

Hb: 9.5 g/dl, Hct: 30.6%, MCV: 83 fL,
WBC: 12,200/mm³, Platelet: 516 000/mm3
Glucose: 20 mg/dl
BUN: 9 mg/dl, Creatinine: 0.19 mg/dl
Uric acid: 2.6 mg/dl
ALP: 23 U/L, AST: 123 U/L (<36)
Creatinine Kinase: 419 IU/L (34-204)
Calcium: 9.8 mg/dl (8.4-10.2), Phosphor: 5.7 mg/dl (2.7-4.5)
Sodium: 132 mEq/l (135-143), Potassium: 4.8 mEq/l (3.1-5.5)
TSH: 1.73 mIU/ml sT4: 1.09 ng/dl, Prolactin: 18.49 ng/ml
FSH: <3 mIU/ml, LH: <0.07 mIU/ml, E2: 29.44 pg

69 aminoacid (prolin) homozygous frameshift mutation: delG209 at the POMC gene

Last visit (At 21 months):

- Body weight: 21 kg (>97 p)
- Height: 85 cm (50-75 p)
- Eyebrows and hair were red

The final steroid treatment dose was 8.9 mg/m2/day.

CONCLUSION

In conclusion, our patient had classic findings of POMC deficiency. However, our options in the treatment of obesity seem to be very limited at the current state of our knowledge.