Somatostatin Experiment in Prohormone Convertase Deficiency

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INTRODUCTION

PC 1/3 is responsible for converting hormones and neuropeptitids which has role energy homeostasis, food intake,glucose metabolism (a-MSH,CART, NPY, AgRP, Orexin, Hypocretin, Ghrelin, insulin, cholecystokinin,GLP-1, GHRH, GnRH, ACTH,TRH) from proforms to active form.

PC 1/3 deficiency's clinical signs are diarrhea that started in the newborn period, obesity,hypoglycemia, multiple pituitary insufficiency i in the infant stage is observed.

CASE

A 4-month-old male patient was admitted to our hospital with diarrhea, dehydration, metabolic acidosis. It was learnt that the diarrhea began when he was 12 days. The patient consulted our department because he had hypoglycemia while receiving intravenous fluid and total parenteral nutrition therapy. In the family history of the patient, the mother and father were first degree cousins.

Physical Examination

Weight: 4590 gr (-2.93 SD), Height: 58.5 cm (-2.13 SD),

BMI: 13.1 kg/m² (-2.78 SD),

Blood Pressure: 55 /30mmHg ,

Thyroid Stage 0, pubertyTanner Stage 1,

Penis lenght:2 cm (<10p), calibration is fine.

The patient's glucose was 44 mg/dl, ketone negative, cortisol and insulin levels were normal but growth hormone was detected to be low.

As a result of the laboratory (Table 1), central hypothyroidism was detected and LT4 therapy was initiated.

fT_4	**0.66**	0.92-1.99	ng/dl
TSH	**4.37**	0.73-8.35	uIU
ACTH	12.9	0-46	pg/ml
Cortisol	16.2	6.2-19.4	ug/dl
FSH	1.85	0.16-4.1	IU/L
LH	<0.1	0.02-0.3	IU/L
T. Testosteron	<0.025	0.003-0.010	ng/mL

Table 1. Laboratory values of Prohormone Convertase Deficiency

The patient's proinsulin: 1300 (3-20pmol/L), Insulin: 12.7 (2.6-25 uIU/mL) was detected. In the genetic analysis of the patient who was diagnosed with Prohormone convertase deficiency, a new mutation of IVS4-2A> G (C.544-2A> G) homozygote was detected. Central hypothyroidism, central adrenal insufficiency and diabetes insipitus were included in the follow up observation of the patient. Somatostatin therapy was started after the catheter was removed from the patient with hypoglycemia due to catheter infection. The patient's hypoglycemia was put under control with somatostatin therapy

CONCLUSION

Hypoglycemia due to proinsulin elevation is seen in cases with Prohormone convertase 1/3 deficiency. Although there are cases of postprandial hypoglycemia in the literature, the corresponding treatment is not shared. Our case hypoglycaemia was prevented by somatostatin therapy. This case is shared because of a new mutation being identified in Prohormone Convertase Deficiency and the first somatostatin experience in the treatment of hypoglycemia.