INTRODUCTION

Costello syndrome is a rare RASopathy that is associated with such characteristics as prenatal overgrowth, postnatal growth retardation, developmental retardation, mental deficiency, coarse face appearance, loose skin on the neck, hands and feet, cardiovascular abnormalities, acanthosis nigricans, deep palmar and plantar lines and a predisposition to various types of cancer. Several endocrine disorders, including growth hormone deficiency, adrenal failure, glucose intolerance, parathyroid adenoma, hyperprolactinemia and hypoglycemia, have been defined in cases with Costello syndrome. In the present report, we describe a patient diagnosed with Costello syndrome accompanied by a clinical picture of hyperinsulinemic hypoglycemia and whose responsive to diazoxide treatment.

CASE

A 36-week-old female patient with a birth weight of 3800 grams showed postnatal growth and developmental retardation, and a physical examination revealed a body weight of 6120 grams, a height of 63 cm, coarse face appearance, and deep palmar and plantar lines, while an echocardiography showed pulmonary valve stenosis. A whole exome sequencing was performed to rule out storage diseases and to confirm the diagnosis of Costello syndrome which was suspected based on clinical findings. The diagnosis of Costello syndrome was confirmed by the presence of the heterozygous missense mutation in HRAS gene alone. A cranial MRI of the patient showed a diffuse thin appearance in corpus callosum. The patient was examined by the endocrinology department at the age of 13 months because of hypoglycemia.

Hyperinsulinism was suspected, since the levels of insulin and c-peptide were elevated when measured while the patient was hypoglycemic, and an exaggerated glucose response was seen in a glucagon test. Blood glucose monitoring indicated episodes of fasting hypoglycemia and postprandial hyperglycemia. Diazoxide of 10 mg/kg/day was initiated in three doses for hyperinsulinemic hypoglycemia, which was resolved, with no new episodes of postprandial hyperglycemia occurring.

CONCLUSION

Very few cases of Costello syndrome accompanied by hyperinsulinemic hypoglycemia have been reported, and the etiology of this condition is yet to be understood, although patients respond well to diazoxide treatment.

REFERENCES