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Multisystemic Severe Form Pseudohypoaldosteronism: Can Gastrostomy be Useful in the Management?

Multisistemik Ağır Form Psödohipoaldosteronizm: Gastrostomi Psödohipoaldosteronizm Yönetiminde Kullanılabilir mi?

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ABSTRACT

A ten-day-old male infant was brought to the hospital with vomiting. On admission, he was hypotonic and had accompanying mild dehydration. The laboratory findings revealed hyponatremia, hyperkalemia, and mild metabolic acidosis. The hormone profile (plasma renin activity: 45 ng/ml/h, aldosterone >20 000 pg/ml, ACTH: 53 pg/ml, 17 OH progesterone: 6.7 ng/ml) revealed pseudohypoaldosteronism (PHA). Oral and IV NaCl, anti-potassium therapy (kayexalate), and low-potassium formula were started. His daily salt requirement was 12 grams. He was not able to take this total salt requirement orally. Respiratory symptoms and diarrhea episodes developed at the age of 3 months. The sodium concentration of sweat was 106 mEq/L. He was therefore diagnosed with multisystemic severe form of type 1 PHA. Severe salt wasting could not be treated. We planned to perform gastrostomy to administer his medications effectively but he died due to a salt-wasting crisis.

Key Words Child, Gastrostomy, Pseudohypoaldosteronism

ÖZET

10 günlük erkek bebek kusma şikayeti ile hastaneye getirildi. Başvuruda hipotonikti ve orta derece dehidratasyonu vardı. Hastanın laboratuvar bulgularında hiponatremi, hiperpotasemi ve orta derecede metabolik asidoz mevcuttu. Hormon profili psödohipoaldosteronizm tanısını desteklemekteydi (plasma renin aktivitesi: 45 ng/ml/h, aldosteron >20 000 pg/ml, ACTH: 53 pg/ml, 17 OH progesteron: 6.7 ng/ml). Oral ve İV NaCl, anti-potasyum tedavi (kayeksalat), düşük potasyumlu mama başlandı. Günlük tuz ihtiyacı 12 gramdı. Olgu tuz ihtiyacının tamamını oral alamamaktaydı. Olgu üç aylık olduğunda solunum semptomları ve diyare epizodları gelişti. Ter sodyum konsantrasyonu 106 mEq/L bulundu. Bu nedenle multisistemik ağır form psödohipoaldosteronizm tip 1 tanısı konuldu. Ağır tuz kaybı karşılanamadı. Tedavilerini etkin verebilmek için gastrostomi planlandı. Ancak olgu tuz kaybı krizi ile kaybedildi.

Anahtar Sözcükler: Çocuk, Gastrostomi, Psödohipoaldosteronizm

INTRODUCTION

Pseudohypoaldosteronism is a rare clinical condition leading to hyponatremia, hyperkalemia and metabolic acidosis. Type 1 PHA is characterized by resistance to aldosterone and is associated with high levels of aldosterone and renin. Salt-wasting crises may be life threatening in infancy. Patients usually require high dose of sodium chloride supplementation with potassium binding resins (1-3). However, giving these high doses can be difficult in clinical practice. These patients may outgrow the illness by 1-2 years of age after appropriate therapy. Here, we report a case diagnosed with type 1 PHA who required gastrostomy. Therefore, we discuss the role of gastrostomy in type 1 PHA management.

CASE REPORT

A 10–day-old male baby presented with poor feeding, vomiting and hypotonia. He was born at term without any complications. There was no consanguinity between the parents. Two elder siblings were also healthy. His birth weight was 3100 grams. His current weight was 2600 grams and height was 49 cm with

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blood pressure 80/45 mmHg, heart rate 138/min, respiratory rate 42/min and body temperature 36.8 oC on admission. Physical examination findings were mild dehydration and hypotonia. External genitalia were normal. Initial biochemical analysis revealed hyponatremia, hyperkalemia and metabolic acidosis with serum Na= 115 mEq/l (N=132-147), serum K= 9.4 mEq/l (N= 3.6-6.1) and serum HCO3= 11.5 mEq/l. His BUN and creatinine levels were increased up to 78 (N:0-25) and 1.22 (N= 0.3- 1.2) mg/dl respectively. Blood glucose and liver function tests were normal. Urine analysis, urine culture, renal ultrasonography results were also unremarkable. However, urinary Na excretion was 106 mmol/L (N= 0-20). Hormonal evaluation results were as follows: plasma renin activity 45 ng/ml/h, aldosterone >20 000 pg/ml, ACTH= 53 pg/ml, 17 OH progesterone: 6.7 ng/ml. Our patient was diagnosed with autosomal recessive form of type 1 PHA and needed peritoneal dialysis at the beginning because of hyperkalemia. After the initial treatment regime he received IV and oral NaCl (IV fluid Na concentration was gradually increased as 75 -100-150-175-250 meq/L), oral NaCl concentration gradually increased as 3-6-9-12 g/day), oral NaHCO3 (1 gram/d), anti-potassium therapy (calcium polystyrene sulfonate/kayexalate), and low potassium formula. His clinical status continued to be stable with acute exacerbation periods. During this period his oral NaCl requirement changed between 6-12 mg/d (78 mEq/kg/d). On follow-up, he was not able to take the total sodium requirements orally and his hospitalization was extended. However, he suffered from vomiting due to gastroesophageal reflux. Anti-reflux therapy and an up-right reflux position were used. The vomiting increased while trying to administer large amounts of salt orally. On the other hand, respiratory symptoms similar to cystic fibrosis developed. Na concentration of sweat was measured as 106 mEq/L. The problems were high sodium requirement, inability to stop IV NaCl, requirement of oral/nasogastric replacement of high amounts of NaCl, frequent respiratory problems, frequent diarrhea, and failure to thrive (his weight was 4400 grams at the age of six months). Nasogastric tube insertion was needed to administer oral NaCl but this route was also unsuccessful. Gastrostomy was required to treat our case effectively. However, he died due to hyperkalemia / salt-wasting crisis while gastrostomy was being planned.

DISCUSSION

Type 1 PHA is a rare inherited disease associated with salt-wasting crisis, life-threatening hypokalemia, metabolic acidosis, dehydration and failure to thrive. It is associated with high levels of plasma renin and aldosterone reflecting resistance to mineralocorticoids (4-6). Type 1 PHA has been classified into two groups. Inactivation of the human mineralocorticoid receptor (MR) due to mutations of the coding gene NRC32 causes autosomal dominant form of type 1 PHA (renal form). Autosomal recessive form of type 1 PHA is the systemic form leading to systemic salt loss including kidneys, colon, sweat and salivary glands (1). Therefore, it causes pulmonary infections due to reduced sodium-dependent fluid absorption (7). Autosomal recessive form of type 1 PHA is caused by inactivating mutations of the ENac (epithelial sodium channel) subunit genes SCNN1A, SCNN1B and SCNN1G (6). Our patient was diagnosed with the systemic form of type 1 PHA because of increased sodium in the sweat and co-existing respiratory problems. However, we were unable to confirm the diagnosis with genetic analysis.

Management of type 1 PHA is very difficult and these cases are prone to life-threatening neonatal-onset salt-wasting crises. Patients diagnosed with the systemic form may require life-long therapy in contrast to the renal form. Treatment options include high-dose sodium chloride, potassium binding resin, IV fluid replacement and also NaHCO3 to correct acidosis (2,3,7-10). Our case was not able to take his medications orally due to respiratory failure, diarrhea and vomiting episodes due to gastrooesophageal reflux. Unfortunately, nasogastric tube insertion was also ineffective. We planned to perform gastrostomy to treat our patient effectively. However, he died because of a sudden salt-wasting crisis before gastrostomy. We did not have the chance to follow-up our case for a long time. We thought that earlier gastrostomy would be beneficial in the management of our case. There are also case reports emphasizing the benefit of gastrostomy in patients with PHA (11,12). Bowden et al (11), reported a boy diagnosed with type 1 PHA with concomitant submucous cleft plate and uvula whose weight gain was satisfactory after gastrostomy.

In conclusion, gastrostomy can be useful in the treatment of type 1 PHA, especially in the systemic form. These patients are prone to feeding difficulties and respiratory problems leading to therapeutic failure. They may benefit from gastrostomy performed at the beginning or in the early period.

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