doi: 10.5262/tndt.2018.3043

Acute Kidney Injury Secondary to Rhabdomyolysis in Case with Gitelman Syndrome

Gitelman Sendromlu Olguda Rabdomiyolize Sekonder Akut Böbrek Hasarı

ABSTRACT

Gitelman syndrome is a genetically transmitted tubulopathy. It is characterized by a disorder in the thiazide-sensitive Na-Cl cotransporter caused by a mutation in the SLC12A3 gene. A case diagnosed with Gitelman syndrome and referred with muscular weakness and cramping complaints due to discontinuing potassium replacement in the follow-up is discussed in this case study. The case was diagnosed with rhabdomyolysis and acute kidney injury secondary to hypokalemia upon determination of a potassium level of 2.14 mEq/L , creatine kinase 27.610 U/L and creatinine 3.09 mg/dL on further examination. Therefore, NaCl 100 cc/h isotonic was administered to the patient in addition to oral and intravenous potassium replacement. The dose of acetazolamide used was 2x250 mg due to the presence of severe metabolic alkalosis. Clinical and laboratory findings were fully restored to normal levels one week after the starting of treatment.

KEY WORDS: Acute kidney injury, Gitelman syndrome, Rhabdomyolysis

ÖZ

Gitelman sendromu genetik olarak geçiş gösteren bir tubulopatidir. Bu SLC12A3 genindeki mutasyon sonucu tiyazid duyarlı Na-CI kotransporter bozukluğu ile karakterize edilir. Bu olgu sunumunda Gitelman sendromu tanısı konulan ve takibinde potasyum replasmanının kesilmesi nedeniyle kas güçsüzlüğü ve kramp şikayetleri ile başvuran bir olgu tartışıldı. Daha ileri incelemelerde 2.14 mEq/L potasyum, kreatin kinaz 27.610 U / L ve kreatinin 3.09 mg / dL belirlenmesiyle rabdomiyoliz ve hipokalemiye sekonder akut böbrek hasarı tanısı kondu. Bu nedenle oral ve intravenöz potasyum replasmanına ek olarak, 100 cc / saat izotonik NaCl verildi. Verilen asetazolamidin dozu, şiddetli metabolik alkaloz nedeniyle 2x250 idi. Tedaviye başlandıktan bir hafta sonra klinik ve laboratuvar bulgular tamamen düzeldi.

ANAHTAR SÖZCÜKLER: Akut böbrek hasarı, Gitelman sendromu, Rabdomiyoliz

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INTRODUCTION

Gitelman syndrome (GS) is a renal tubular disease presenting with electrolyte imbalance (1). It is induced by the defect in the Na-Cl cotransporter in distal tubules. It manifests itself with hypokalemia without hypertension, hypomagnesemia, hypercalciuria and metabolic alkalosis (2). Patients may commonly present with muscular weakness, spasm and cramping, and also acute kidney injury (AKI) secondary to rhabdomyolysis in case of long-term severe hypokalemia. Case reports on hypokalemia, rhabdomyolysis and AKI secondary to GS are limited in the literature. A case followed-up with a GS diagnosis, referred with muscular weakness and cramping complaints and found to have rhabdomyolysis and AKI secondary to hypokalemia is discussed in the current case report.

CASE REPORT

A 32-year old male patient presented to the emergency service with muscular weakness and cramping for two days. The history revealed that he had a previous

Received : 03.11.2017
Accepted : 14.12.2017

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diagnosis of GS 12 years prior. He was therefore on potassium citrate treatment which he had discontinued during the previous week with no other medication. The patient did not smoke or drink alcohol. The family history was negative. Physical examination revealed palpation and sensitivity in the extremities with no other abnormal examination findings. Vital findings were temperature 36 °C, blood pressure 110/60 mmHg, heart rate 84 bpm, and respiratory rate 12/min. The laboratory tests of the patient revealed urea: 31 mg/dL, creatinine: 3.09 mg/dL, potassium: 2.14 mEq/L, pH: 7.56 and HCO_3: 59 mmol/L. The patient was cramping and was found to have a creatine cinase (CK) level of 27.610 U/L, aspartate aminotransferase 258 IU, and aldolase 240 ng/ml (reference: 0 – 10 ng/ml) with normal findings on thyroid function tests. Other electrolyte levels including calcium and routine laboratory parameters were determined to be normal. Kidney dimensions and parenchymal echogenicity were determined to be normal without hydronephrosis in the urinary system of the patient imaged by ultrasound. Renal artery Doppler ultrasound findings were normal. It was learned that the kidney function tests of the patient had given normal results one month before. There was no potential cause of dehydration such as nephrotoxic agent intake, contract exposure, or diarrhea-vomiting. Thus, the findings suggested that the patient had rhabdomyolysis and AKI secondary to hypokalemia. Therefore, NaCl 100 cc/h isotonic was administered to the patient in addition to oral and intravenous potassium replacement. The dose of acetazolamide used was 2x250 due to presence of severe metabolic alkalosis. During follow-up one week after the beginning of treatment, test findings were potassium: 3.42 mEq/L, pH: 7.42, HCO_3: 24.5 mmol/L, CK 272 U/L and creatinine 1.16 mg/dL. The symptoms of patient were fully recovered. Acetazolamide treatment was therefore stopped, potassium citrate one tablet od was prescribed and the patient was discharged with further follow-up to be performed at the outpatient clinic.

DISCUSSION

Gitelman syndrome is a rare tubulopathy transmitted as an autosomal recessive trait. It is characterized by a disorder in the thiazide-sensitive Na-Cl cotransporter caused by a mutation in the SLC12A3 gene (3). Patients may present with hypokalemia, hypomagnesemia and metabolic alkalosis-associated muscular weakness and cramping (2). Patients are usually diagnosed with GS during puberty and adulthood.

Rhabdomyolysis is induced by several factors such as muscular trauma, alcohol consumption, infection, enzyme deficiencies and endocrine disorders. Hypokalemia, which is an electrolyte disorder, is among these reasons (4). Rhabdomyolysis associated with hypokalemia is an expected condition in GS patients. The literature widely reports hypokalemia and rhabdomyolysis secondary to GS. However, a literature review resulted in a number of pediatric cases presenting with GS => hypokalemia => rhabdomyolysis => AKI. Kumagai et al. (5) reported in their case that a 13-year-old girl was referred with muscular weakness and cramping. Further examination led to the establishment of the diagnosis of GS and hypokalemia => rhabdomyolysis => AKI secondary to GS. The patient who did not have severe metabolic alkalosis was administered oral and intravenous potassium and hydration treatment. Laboratory and clinical findings of the patient were restored to normal levels on the 10th day of treatment. In contrast to the case mentioned above, our case had already been diagnosed with GS 12 years prior to his referral, and rhabdomyolysis and AKI had developed secondary to severe hypokalemia induced by discontinuing the potassium treatment he was on. While the case reported by Kumagai et al. (5) was in the pediatric age group, our case is in the adult age group. Therefore, our case is the first case to be reported in the adult age group.

It has been demonstrated that the level of CK is correlated to the risk of AKI development in rhabdomyolysis cases. Talving et al. determined in their study that a CK level >3000 IU/L is an independent risk factor for the development of AKI (6). Rodríguez et al. determined in their study that a CK level >5000 IU/L is associated with AKI (7). The CK level was determined as 27.610 U/L in the current patient, and it is significant in respect to AKI in accordance with the mentioned studies.

In conclusion, it should be remembered that rhabdomyolysis secondary to severe hypokalemia which induces AKI may develop in GS patients.

REFERENCES

1. Knoers NV, Levtchenko EN: Gitelman syndrome. Orphanet J Rare Dis 2008;3:22
2. Al Shibli A, Narchi H: Bartter and Gitelman syndromes: Spectrum of clinical manifestations caused by different mutations. World J Methodol 2015;5:55-61
3. Nakhoul F, Nakhoul N, Dorman E, Berger L, Skorecki K, Magen D: Gitelman's syndrome: A pathophysiological and clinical update. Endocrine 2012;41:53-57
4. Luzardo L, Silvariño R, Boggia J, Noboa O, Gadola L: Severe hypokalemic rhabdomyolysis. Report of six cases. Rev Med Chil 2014;142:651-655
5. Kumagai H, Matsumoto S, Nozu K: Hypokalemic rhabdomyolysis in a child with Gitelman's syndrome. Pediatr Nephrol 2010;25:953-955
6. Talving P, Karamanos E, Skiada D, Lam L, Teixeira PG, Inaba K, Johnson J, Demetriades D: Relationship of creatine kinase elevation and acute kidney injury in pediatric trauma patients. J Trauma Acute Care Surg 2013;74:912-916
7. Rodríguez E, Soler MJ, Rap O, Barrios C, Orfila MA, Pascual J: Risk factors for acute kidney injury in severe rhabdomyolysis. PLoS One 2013;8:e82992