Mixed diabetic ketoacidosis and hyperglycemic hyperosmolarity in a girl with nephronophthisis 4 presenting with rhabdomyolysis and pancreatitis

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To the editor,

Diabetic ketoacidosis (DKA) and hyperglycemic hyperosmolar state (HHS) are the 2 most severe acute hyperglycemic crises of diabetes.¹ Only a few pediatric cases of mixed HHS and DKA have been reported.² The combination of DKA with HHS is associated with increased mortality and treatment difficulty.³ We present a case report of a 16-year-old female who had no medical problems except obesity and who was admitted to the hospital with severe DKA with HHS as the first manifestation of diabetes mellitus. She presented with severe metabolic acidosis, renal failure, rhabdomyolysis, pancreatitis, and coccyx sore. Conventional treatment of DKA and renal replacement therapy resulted in quick recovery from DKA and the metabolic profile, but the hospital stay was prolonged due to continued pancreatitis and exacerbation of a pressure sore. After 15 days of fasting, her pancreatitis improved, and she was able to eat. However, the hospital stay was extended to 70 days for management of necrotic changes of the coccyx sore. After discharge, her renal function has not completely recovered. Her older sister had chronic renal failure, based on which whole exome sequencing was performed to determine the genetic cause of renal disease. As a result, a homozygous mutation c.2304+1G>A in NPHP4 was identified. According to the ACMG 2015 guideline, this mutation has demonstrated pathogenic variants associated with nephrolithiasis (NPHP). NPHP is one of the most common causes of chronic renal failure in children, responsible for 2.4% to 15% of patients with end-stage renal disease in this population.⁴ To date, there are no reports of diabetes in pediatric patients with NPHP. One-half of patients with NPHP develop an extrarenal phenotype such as retinal dystrophy, cerebellar hypoplasia, mental retardation, situs inversus, polydactyly, and hepatic cysts.⁵ Currently, her renal function has deteriorated and is accompanied by anemia and metabolic acidosis, possibly due to the NPHP4 mutation; no other extrarenal symptoms are evident.

This is the first report of mixed DKA and HHS with various complications in a patient with nephrolithiasis 4. Though acute renal failure is not uncommon in DKA/HHS, genetic testing should be considered in cases where renal function is not consistently restored or there is a family history of chronic renal failure to identify any other underlying causes.

Notes

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