Beckwith–Wiedemann syndrome and recurrent bilateral renal calculi

Sir,

Patients with Beckwith–Wiedemann syndrome, a congenital condition classified as an overgrowth syndrome, have a multitude of clinical manifestations including large body size, large organs including abnormally large kidneys.[1] Medullary sponge kidney also occurs more frequently in patients with this congenital disorder. As we described in a recently published article,[2] in the current era, medullary sponge kidney can be easily missed since intravenous urography is not utilized as often as it used to for the investigations for kidney stones. Thus, it is important to recognize the characteristic of medullary sponge kidney in computed tomography (CT) scan. Hereby, we present a 70-year-old male with Beckwith–Wiedemann syndrome and recurrent kidney stones despite being on chlorthalidone. Physical examination showed crossed hemihypertrophy; one arm and the contralateral leg are larger than their counterparts. The analysis of the passed stones was 100% calcium phosphate (apatite). His blood test revealed normal creatinine of 1.1 mg/dL. His urine pH was 7.5. A 24-h urine was supersaturated with hydroxyapatite. Otherwise, the patient also had hypocitraturia (120 mg/sec). Abdomen CT without contrast showed extensive bilateral medullary nephrocalcinosis and bilateral kidney stones. The largest stone measures 1.4 cm × 1.0 cm in the lower pole of the right kidney [Figure 1]. Dual energy CT characterization blue color coding showed that the stone element was nonuric acid in composition [Figure 2]. Thus, the patient was diagnosed with Beckwith–Wiedemann syndrome and medullary sponge kidney with bilateral nephrocalcinosis/kidney stones. Chlorthalidone and potassium citrate were started. At 1-year follow-up, his stone passages have diminished considerably.

Physicians should be aware of the manifestations of medullary sponge kidney and nephrocalcinosis in patients with Beckwith–Wiedemann syndrome. In addition to nephrocalcinosis, renal cysts, recurrent pain associated with infundibular stenoses, nephroblastomatosis, and Wilms tumor have also been reported.[3] As shown in our case presentation, patients with medullary sponge kidney usually form calcium phosphate in their stones due to their increased urinary pH.[1,4] Although hypercalciuria was not found in our case, hypercalciuria has also been described in patients with Beckwith–Wiedemann syndrome.[5]

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Wisit Cheungpasitporn, Stephen B. Erickson

Division of Nephrology and Hypertension, Mayo Clinic, Rochester, MN, USA

Address for correspondence:
Dr. Wisit Cheungpasitporn,
Mayo Clinic, 200 First Street SW, Rochester, MN 55905, USA.
E-mail: wcheungpasitporn@gmail.com

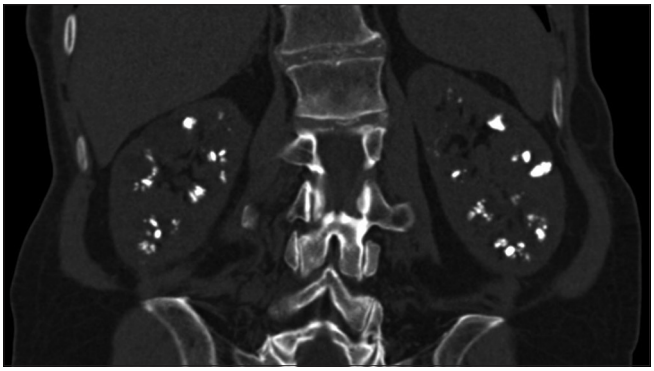

Figure 1: Abdomen computed tomography without contrast demonstrated numerous small bilateral nonobstructive renal calculi at papillary tips suggestive of medullary sponge kidneys

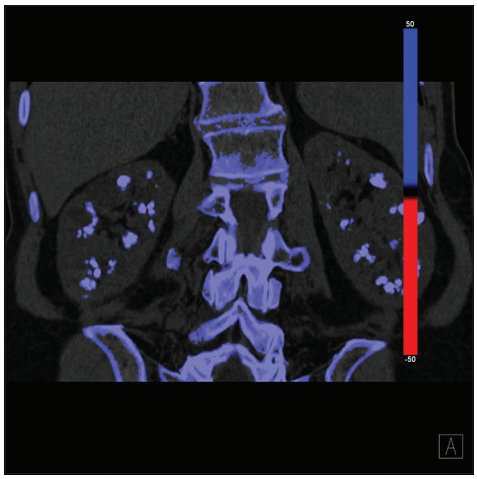

Figure 2: Dual energy computed tomography characterization blue color coding indicated that the stone material was nonuric acid in composition

REFERENCES

1. Weksberg R, Shuman C, Beckwith JB. Beckwith-Wiedemann syndrome. Eur J Hum Genet 2010;18:8-14.
2. Cheungpasitporn W, Erickson SB. Medullary sponge kidneys and the use of dual-energy computed tomography. Urol Ann 2015;7:129.
3. Kulkarni R, Wolf JS Jr., Padiyar N, Zuckerman L, Gera R, Scott-Emuakpor AB. Severe intrarenal fibrosis, infundibular stenosis, renal cysts, and persistent perilobar nephrogenic rests in a patient with Beckwith-Wiedemann syndrome 27 years after diffuse nephroblastomatosis and Wilms tumor: Natural progression or a consequence of treatment? J Pediatr Hematol Oncol 2002;24:389-93.
4. Cheungpasitporn W, Thongprayoon C, Brabec BA, Kittanamongkolchai W, Erickson SB. Outcomes of living kidney donors with medullary sponge kidney. Clin Kidney J 2015. [Epub ahead of print].
5. Goldman M, Shuman C, Weksberg R, Rosenblum ND. Hypercalciuria in Beckwith-Wiedemann syndrome. J Pediatr 2003;142:206-8.

Access this article online	
Quick Response Code:	Website: www.urologyannals.com
	DOI: 10.4103/0974-7796.198837

How to cite this article: Cheungpasitporn W, Erickson SB. Beckwith–Wiedemann syndrome and recurrent bilateral renal calculi. Urol Ann 2017;9:113-4.