Open Journal of
Clinical & Medical
Case Reports

Volume 5 (2019)
Issue 11

ISSN: 2379-1039

Case of the hidden syndrome: A rare constellation of findings

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Abstract

A 56-year old male with presented with a chief complaint of pain in the right flank area, which raised suspicion for nephrolithiasis. Nephrolithiasis is the most common chronic kidney condition, after hypertension. After further investigation, a CT scan of the abdomen and pelvis utilizing renal protocol showed 4x stones in the right kidney, with a non-obstructing kidney noted to be malrotated. Upon general examination, the patient is a morbid obese male with profound hypogonadism who also complains of erectile dysfunction. The evaluation of his laboratory values demonstrated low LH, FSH, and normal testosterone levels. Therefore, a pituitary etiology is suspected, however, this patient's MRI did not reveal any significant findings explaining the elevated prolactin levels. To our knowledge, we present the first case report with this specific rare constellation of impressive physical findings which included malrotation of the right kidney, renal stones, urachal remnant, umbilical hernia, profound hypogonadism, due to pituitary failure, a normal sella turcica on MRI, and morbid obesity. The authors believe that these findings may be concomitant with an undiscovered syndrome with an associated genetic origin.

Keywords

urology; nephrolithiasis; hypogonadism; stone disease; renal calculi

Introduction

Nephrolithiasis is the most common chronic kidney condition, after hypertension. The initial presentation of nephrolithiasis is often with renal colic, which is defined as severe pain caused by stone passage. Renal colic may be triggered by movement of a stone form the renal pelvis into the ureter, which leads to ureteral spasms and possible obstruction. The patient may present with pain which initially begins in the flank area and progresses downward and anteriorly into the genital region as the stone moves down the ureter. The pain experienced is not typically aggravated or alleviated by positional changes and may be accompanied by nausea and vomiting. This gives the classical presentation of a patient who is constantly moving in order to find a comfortable position, as opposed to the pain experienced in peritonitis where the patient lies still and is very protective of their abdomen [1].

Many dietary factors such as calcium and fluid intake have a major role in the formation of urinary stones [2]. Over the last few decades, there have been great advancements in minimally invasive techniques. Currently, treatment options include extracorporeal shock wave lithotripsy (ESWL), percutaneous nephrolithotomy, retrograde intrarenal surgery, and laparoscopic ureterolithotomy [3]. ESWL, which uses sound waves to fragment stones into small pieces that can be easily passed, is effective for most stones less than 2 cm in size (Worcester). Medical therapy is a watchful waiting approach for treating urethral calculi and can be used successfully for a considerable number of patients [4]. Alpha-adrenoreceptor antagonists, calcium channel blockers, and phosphodiesterase-5 inhibitors are believed to act by relaxing the ureteral smooth muscle to reduce ureteral contractions, inhibiting peristalsis and aiding in the elimination of stones. This medical management also reduces the frequency of colic pain [5].

Consequently, while surgical modalities are still considered the mainstay of treatment for urolithiasis, medical expulsion therapy has recently emerged as an alternative treatment modality for the management of distal ureteric stones [6].

Case Presentation

A 56-year old male with presented with a chief complaint of pain in the right flank area. Three years ago, he had an ESWL in Cuba. Patient had this pain for three months, and he also complains of urine frequency and erectile dysfunction. At the time, the only medications the patient was taking was ibuprofen, and was taking no medication for his urinary symptoms. The patient's past medical history is significant for arthritis. Previous surgical history includes a surgery for an umbilical hernia and an appendectomy. The patient's family history is significant for kidney disease in both his brother and father, it should also be noted that his mother had stomach cancer. Upon his review of systems there were no significant findings. The patient appeared well-developed, well-nourished, well-groomed, was in no apparent acute distress, and was oriented to time, place, and person.

A CT scan of the abdomen and pelvis utilizing renal protocol showed 4x stones in the right kidney, with a non-obstructing kidney noted to be malrotated. The kidneys were of normal size with the right kidney malrotated antero-posteriorly. The lower and mid pole moiety on the right contained calculi measuring 5 mm in the lower pole. The mid-pole had a pair of calculi measuring 8 x 5 and 7 x 5 mm adjacent each other. There was an additional calculus measuring 6 x 4 mm. the collecting system was not distended and no calculi were identified along the right ureter. The left kidney did not demonstrate any abnormalities. In addition, a urachal remnant and an umbilical hernia were both present. It should also be noted that the patient had relatively advanced vascular calcifications for his age, particularly in the aorta. The prostate was relatively small with normal appearing seminal vesicles. Multiple distal colonic diverticula were present without any evidence of diverticulitis. The lung bases demonstrated coarse interstitial markings bilaterally. The heart was normal size with vascular calcifications. The liver was borderline in size and demonstrated minimal to mild fatty infiltration. It should also be noted that the adrenal glands, pancreas, gallbladder, and spleen all appeared normal. The patient's testosterone level was profoundly low at 102 ng/dl. LH and FSH levels were also profoundly low and his prolactin level was elevated. A prostate-specific antigen level (PSA)

was noted to be at a level of 0.3 ng/ml.

A MRI of the brain was performed without and with IV contrast due to the patient's elevated prolactin levels. Images demonstrated no mass or abnormal enhancement. No parenchymal signal abnormalities were noted. The sella turcica demonstrated no evidence of sellar or suprasellar mass. The pituitary gland was in normal size and signal with normal enhancement and no discrete lesions.

Upon general examination, the patient is a morbid obese male with profound hypogonadism who also complains of erectile dysfunction. The evaluation of his laboratory values demonstrated low LH, FSH, and normal testosterone levels. There was no pituitary adenoma to account for the abnormal testosterone axis labs. Therefore, this is a pituitary failure and could be a micro-adenoma. Metabolic syndrome and the patient's obesity was ruled out as a potential etiology for his hypogonadism, otherwise there would have been elevated FSH and LH levels. The current case presents an interesting gathering of findings including malrotation of the right kidney, renal stones, urachal remnant, umbilical hernia, profound hypogonadism due to pituitary failure, a normal sella turcica on MRI, and morbid obesity.

Treatment

The patient was first prescribed ibuprofen 800 mg three times daily with meals following his initially evaluation with his presenting chief complaint of renal colic pain of 3 months duration. He was asked to follow up with the physician 2 weeks later following his CT scan of the abdomen and pelvis. Following assessment of the patient's imaging studies, the decision was made with the patient to perform extracorporeal shock wave lithotripsy. The patient was instructed to continue self-injection with testosterone cyprionate 200 mg/ml which was prescribed to him 7 months ago. The patient has had an uneventful post-treatment course following the ESWL with no recurrent bouts of renal colic or hematuria.

Discussion/Conclusion

We present an unusual case of a patient presenting with multiple kidney stones, who upon further evaluation has a collection of impressive physical and laboratory findings as well a significant family history that raises suspicion and warrants further investigation. The prevalence of nephrolithiasis in the United States is 8.8%, affecting 1 of 11 individuals, with a significantly growing incidence [7]. Diabetes, obesity, hypertension, and metabolic syndrome are interrelated clinical problems that have all individually been associated with an increased risk of nephrolithiasis. Otunctemur et al. found that that men with testosterone concentrations (<2.85 ng/mL) had three time higher odds of kidney stones [8].

Metabolic syndrome has a wide range of long-term complications, including non-alcoholic fatty liver disease, polycystic ovarian syndrome, obstructive sleep apnea, chronic renal disease, and hypogonadism [5]. While hypogonadism may be seen in associated with nephrolithiasis, the presenting findings in the current case report rules out metabolic syndrome as a potential etiology. If metabolic syndrome was the cause of the patient's hypogonadism, there would have been elevated FSH and LH levels.

Hyperprolactinemia is a very common condition frequently associated with the use of certain me-

dications [12]. The investigation of hyperprolactinemia in people using antipsychotic medications is complicated because of the well-known association between medications and hyperprolactinaemia [13]. The current case presents a patient with elevated prolactin levels with no associated use of hyperprolactinemia-inducing medication. Therefore, a pituitary etiology is suspected, however, this patient's MRI did not reveal any significant findings explaining the elevated prolactin levels.

Urachal remnants can present as one of four primary recognized pathologies; patent urachus, urachal sinus, vesicourachal diverticulum, and urachal cyst. Patent urachus involves free communication between the bladder and the umbilicus, and presents with urine leakage through the umbilicus or occasionally with a urinary tract infection [9]. Though urachal anomalies are rare, the clinician must be highly suspicious as urachal cystic tissue can potential lead to bladder adenocarcinomas [11]. Since this is considered a rare disorder there are frequent misdiagnoses [10]. It is evident that urachal anomalies should be considered in the differential of abdominal pain to ensure timely and appropriate management. It is the rarity of this condition which gives the current report such clinical significance. In addition, the patent urachus in this patient could be associated with an underlying syndrome which has yet to have an established diagnosis.

The current available literature fails to describe any cases with a similar collection of associated findings seen in a single patient with a family history significant for kidney disease in two subsequent generations. To our knowledge, we present the first case report with this specific rare constellation of impressive physical findings which may be concomitant with an undiscovered syndrome with an associated genetic origin.

Acknowledgement

The authors wish to thank Florida International University College of Medicine for giving permission to allow the study to be conducted.

Author contributions

Author Daniel Indorato, who is the principle author in this study, was responsible for gathering the patient's information and writing the paper. The principle author made sure that informed consent was obtained before the study was conducted. Dr. Robert Kester was responsible was obtaining informed consent as well. He was also responsible for treating the patient as well as giving the principle author all of the appropriate labs, imaging, and patient information that was necessary for the case report to be written.

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Manuscript Information: Received: June 17, 2019; Accepted: July 26, 2019; Published: July 31, 2019

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Citation: Indorato D, Kester R. Case of the hidden syndrome: A rare constellation of findings. Open J Clin Med Case Rep. 2019; 1569.

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