

Review Article

Urolithiasis and Nephrocalcinosis in Medullary Sponge Kidney - Urologist's Perspective

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Abstract

Medullary Sponge Kidney (MSK) is a rare renal malformation associated with Nephrocalcinosis and renal stone with urinary acidification and concentration defects. The exact cause of medullary sponge kidney is not known. The hallmark of MSK is cystic dilation and ectasia of the terminal collecting ducts. It manifests only later in adult life, especially during 3rd or 4th decade of life.

The very fact that MSK is associated with various malformative conditions supports the viewpoint that MSK by itself could be a developmental disorder. Further molecular studies are needed to confirm the hypothesis. Most MSK cases are asymptomatic and do not present with any clinical manifestation until the 3rd decade of life. MSK may be unilateral or bilateral and may involve single or multiple pyramids. 70% are bilateral at the time of presentation.

Medullary sponge kidney is a disease diagnosed by excretory urography. With better axial imaging facilities available these days and with an increasing tendency amongst the urologists to avoid Intravenous urography these days, it looks as though the number of MSKs diagnosed these days is on the decline. MSK is a disease that deserves more attention and awareness. MSK is a disease that deserves more attention and awareness. The diagnosis by CT scanning is unreliable and the condition is easily over diagnosed or diagnosis by excretory urography is so scantily done and the disease is easily under diagnosed. A high index of clinical suspicion is necessary in order to avoid an under or over diagnosis of this rare entity.

Keywords: Urolithiasis; Medullary sponge kidney; Nephrocalcinosis

Introduction

The term “Nephrocalcinosis” by itself has diverse meanings and clinical implications. By strict terms, this terminology refers to deposition of calcium salts within the renal parenchyma, but there is a considerable overlap in the usage of this terminology by the urologists, between the calcifications that one finds within the renal parenchyma and the stones that are found in the collecting system. It is observed that a varied group of diseases can produce this appearance including those that cause systemic metabolic alterations of hypercalcemia and hypercalciuria. Even though Albright had coined the term nephrocalcinosis for reporting those radiologically demonstrable diffuse renal calcifications in the renal parenchyma, the term nephrocalcinosis has now been extended to calcium deposition in other conditions like type 1 renal tubular acidosis and medullary sponge kidney as well [1].

Nephrocalcinosis by definition refers to deposition of calcium salts within the renal parenchyma. Based on its location it can be classified as either cortical or medullary nephrocalcinosis. Cortical nephrocalcinosis occurs usually as a consequence of chronic glomerulitis or renal cortical necrosis secondary to vascular

insult [2]. On the other hand, medullary nephrocalcinosis occurs secondary to medullary sponge kidney, renal tubular acidosis and hyperparathyroidism. Such calculi may also form due to stasis of urine in collecting ducts and hypercalciuria [3]. The purpose of this current report is to share the insight gained into this clinically important distinction and to highlight the various urological issues related to diagnosing and treating stones in patients with medullary sponge kidney.

Etiopathogenesis

Medullary Sponge Kidney (MSK) also known as Cacchi-Ricci disease or Precalyceal canalicular ectasia is a rare renal malformation associated with a high risk of nephrocalcinosis and renal stones, with urinary acidification and concentration defects, cystic anomalies of pre calyceal ducts and a moderate risk of developing urinary infections and renal failure. It may affect one or both kidneys. The exact cause of medullary sponge kidney is not known. It mostly occurs sporadically, but familial cases have been reported [4].

The hallmark of MSK is cystic dilation and ectasia of the terminal collecting ducts. Like in distal renal tubular acidosis, nephrocalcinosis is common. These two conditions may be difficult to distinguish with plain X-ray KUB alone. Intravenous pyelography is much more useful in demonstrating the cystic dilation of the collecting ducts and the stones lying within the dilated tubules. Such patients also have hypercalciuria that can further increase the risk of stone formation.

MSK was first identified in 1939 by Lenarduzzi. It manifests only later in adult life, especially during 3rd or 4th decade of life. MSK is an anatomical abnormality of renal unit development and is associated with certain other changes that are of interest to the urologist. MSK occurs with an incidence of 1 in 5,000 to 1 in 200,005 [5]. Herewith, we discuss the various aspects of MSK and its presentation and treatment required.

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Normal renal units develop from the metanephric blastema and any abnormality arising leads to malformations and anomalies. MSK arises due to disruption of the ureteric bud and metanephric blastema interface. Because of this disruption, the calyceal ends of the ducts of Bellini do not form proper units and end up forming dilated cystic segments and hence the disease. The main pathology seems to be cystic dilatation of the collecting ducts of the nephron, leading to cyst formation, found exclusively in the medullary portion and sparing the cortex. These cysts at the collecting ducts lead to defective secretion and reabsorption causing defective tubular acidification and concentration abilities. These cystic portions along the nephron pathway are also portions of urinary stasis and favor stone formation. They also have reduced urinary levels of inhibitors (citrate and magnesium). Thus many compounding factors are in thought to be contributory to stone formation in patients with MSK [6,7].

Molecular Basis

The very fact that MSK is associated with various malformative conditions supports the viewpoint that MSK by itself could be a developmental disorder. Choyke et al. [8] had identified a 13% association in patients with Beckwith Wiedmann syndrome. Gambaro et al. [9] identified a 10% prevalence of malformations in a cohort of MSK patients. Kerr et al. [10] noted a 70% prevalence of MSK in patients with congenital hepatic fibrosis and Caroli's syndrome. Apart from these strong associations, it may also be possible that the differentiative abnormality occurs throughout both kidneys, only in one kidney, or even in just a few papillae [11]. Further molecular studies are needed to confirm the hypothesis.

Clinical Features

Most MSK cases are asymptomatic and do not present with any clinical manifestation until the 3rd decade of life, where they may present with pain as the only major symptom. MSK may also present with stones, UTI and hematuria. MSK may present with multiple stone formations as medullary nephrocalcinosis and it may appear with certain specific characteristics on various radiological imaging. MSK patients have a 60% lifetime risk of stone formation (calcium oxalate and calcium phosphate stones). MSK maybe unilateral or bilateral and may involve single or multiple pyramids. 70% are bilateral at the time of presentation.

Radiology Imaging

Medullary sponge kidney is a disease diagnosed by excretory urography. With better axial imaging facilities available these days and with an increasing tendency amongst the urologists to do Intravenous urography these days, it looks as though the number of MSKs diagnosed these days is on the decline.

Excretory urography has certain typical findings that can strongly confirm the diagnosis of MSK. There is accumulation of contrast in the dilated portion of the collecting ducts that give rise to the "brush appearance". These are triangular shaped opacities with their base facing laterally, giving the characteristic "bouquet of flowers" appearance. As ultrasound, spiral computed tomography, nuclear magnetic resonance does not use contrast media, and fewer cases of MSK are being diagnosed. Ultrasound simply shows nonspecific signs of hyperechoic medulla due to nephrocalcinosis that does not characterize whether it is due to MSK or any other condition [12,13]. Similarly, computed tomography also would suggest the possibility of MSK (Figure 1), but cannot categorically help us make a diagnosis of the disease [14]. Multiphasic helical computed tomography, which is extensively indicated in evaluation of microscopic hematuria, has

been reported to be more sensitive than urography [15]. It is even more helpful in patients with dropped down stone in the upper ureter and presenting with acute colic (Figure 2). In such instances, excretory urography may not be beneficial, if the pelvi calyceal system is non-visualized. Similarly, nuclear magnetic resonance imaging also does not seem to be sensitive enough to identify the typical signs of MSK [16]. However Magnetic resonance imaging and urogram have been found to be superior in making a diagnosis of MSK [17]. Hence, Gambaro et al. [18] had concluded that, with lesser and lesser number of excretory urography being done nowadays, there is a substantial likelihood of this renal condition being a forgotten entity in the near future.

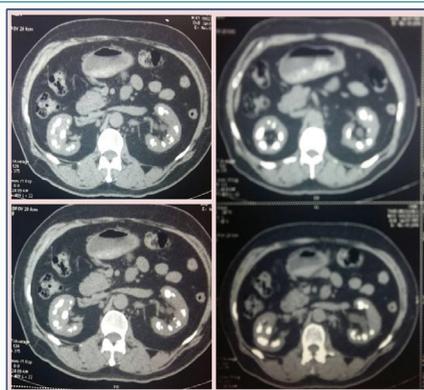


Figure 1: Cross section CT scan showing nephrocalcinosis in medullary sponge kidney.



Figure 2: Coronal section of CT scan showing renal and upper ureteric stones (arrow mark).

Management

MSK is a multifaceted and poorly understood disease. It can manifest uniquely from patient to patient. In that respect what works for one person may not be appropriate for another and treatment strategies should thus be titrated on a patient to patient basis.

Management of MSK largely depends on the type of presentation. It may be due to multiple stone formation or recurrent UTIs or hematuria. Most of the times, MSK needs only passive surveillance with regular fixed follow up with the physician regarding the renal pathology or when symptoms arise. This creates a management dilemma in the minds of the treating urologists regarding when to offer definitive treatment.

It is now generally accepted that asymptomatic patients be regularly screened for hypercalciuria and bacteriuria. Hypercalciuria is treated with salt restriction and plenty of fluids. In addition thiazide therapy may be useful in bringing down the levels of hypercalciuria.

If the urine is infected, it should be treated aggressively with culture specific appropriate antibiotics until urine becomes sterile [19]. Extra corporeal Shock Wave Lithotripsy (ESWL) can be tried whenever the stones block the collecting system and causes colic and hydronephrosis [20]. Regular follow-up is important in such patients.

They may mostly require only conservative management for that particular episode and followed by adequate hydration to prevent UTIs and recurrent stone formations. Such an observant management is more than sufficient most of the time and rarely only cause's renal unit functional impairment. Endourological ureteroscopic intervention is required only when the stone has down migrated into the ureter and causing obstruction and colic. In case of development of large volume stones necessitating removal, percutaneous approaches have been tried in several studies with good results for stone clearance, although it is technically demanding [21].

Conclusion

MSK is a disease that deserves more attention and awareness. Further research efforts are required to clarify its etiology and optimize treatment strategies. The diagnosis by CT scanning is unreliable and the condition is easily over diagnosed or diagnosis by excretory urography is so scantily done and the disease is easily under diagnosed, leading to all possible confusions in diagnosis and management. Confirmation with flexible upper tract endoscopy can make a definitive diagnosis in patients suspected to have MSK and can be diagnostic and many a time potentially therapeutic in terms of stone removal. It is prudent to consider urographic phase imaging either with excretory urography or CT urogram to confirm the diagnosis in cases where upper tract endoscopy is performed. Various other conditions including ductal plugging can often mislead in making a proper diagnosis. A high index of clinical suspicion is necessary in order to avoid an under or over diagnosis of this rare entity.

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