Medullary sponge kidney and isolated hemihyperplasia

P. S. Priyamvada, S. Parameswaran, M. Sandeep, V. Shankar, R. P. Swaminathan[1]

Departments of Nephrology, and [1]Medicine, Jawaharlal Institute of Postgraduate Medical Education and Research, Puducherry, India

ABSTRACT

The term hemihyperplasia refers to an enlargement of body parts beyond the normal asymmetry. Hemihyperplasia can be isolated or associated with various well-described malformation syndromes. Medullary sponge kidney (MSK) has been described with isolated and syndromic hemihyperplasia; the actual prevalence is not known The hemi hypertrophy can be so subtle that it may be easily overlooked. MSK need not be limited to the side of hemihyperplasia – most often it is bilateral. Around 33 cases has been reported from different parts of the world of which 15 cases are isolated hemi hyperplasia (IHH), the remaining occurring in the context of various malformation syndromes So far only one case has been reported from India. We report a case of IHH involving right side of the body, recurrent renal stones, incomplete distal renal tubular acidosis hypercalciuria and imaging showing bilateral MSKs.

Key words: Hemihyperplasia, medullary sponge kidney, nephrolithiasis

Introduction

Medullary sponge kidney (MSK) (Lenarduzzi-Cacchi-Ricci disease) is a relatively uncommon benign developmental malformation which remains clinically silent unless complicated by renal stones or recurrent urinary tract infections.[1] It is rarely associated with malformation syndromes like Isolated hemi hyperplasia (IHH), Beckwith-Weidman syndrome and Carolis disease. The extent of the body part hyperplasia also varies from patient to patient; there are no consensus criteria for the diagnosis of IHH; usually it is subtle that it may be easily overlooked. So far around 15 cases of IHH have been reported from different parts of the world.[2]

Case Report

The present case report is about a 33-year-old female patient who was referred to our department for evaluation of recurrent renal stones. She had recurrent episodes of flank pain since 2002, with one session of extracorporeal shock wave lithotripsy (ESWL) done 8 years back. She continued to experience flank pain after ESWL with history of passing stones. On examination, she had overgrowth of the entire right half of the body. The upper limbs measured 57 cm on the right and 54 cm on the left; lower limb measured 90 cm on right and 85 cm on the left; right foot was larger than the left by 3 cm [Figure 1]. The right half of the trunk was also larger than left; the discrepancy in the length of lower limb resulted in scoliosis with pelvic tilt. Patient recalled of having limping gait from early child hood.

On evaluation, she was found to have a glomerular filtration rate of 114 ml/min/1.73 m^2 of body surface area, urine sediment was non-remarkable except for pyuria; urine cultures did not grow any organisms. Her serum sodium, potassium, calcium and phosphorus

Address for correspondence:
Dr. P. S. Priyamvada, Department of Nephrology,
Jawaharlal Institute of Postgraduate Medical Education and Research,
Puducherry - 605 006, India.
E-mail: priyamvadaps@gmail.com

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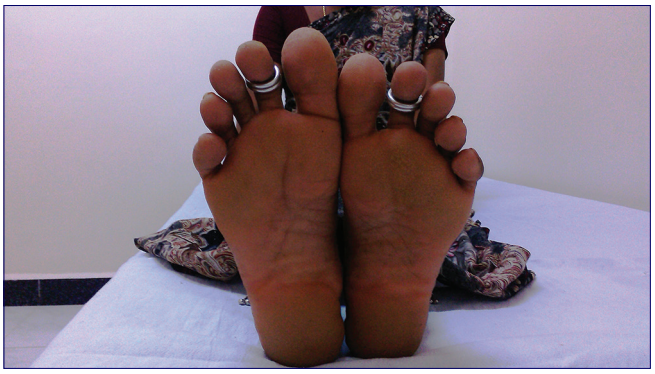

Figure 1: The difference in the size of foot. Right foot is larger than left by 3 cm

were normal and intact. Parathyroid hormone was also in the normal range. A 24 h urine collection showed hypercalciuria (560 mg in 24 h), citrate excretion in the low normal range (265 mg in 24 h) and potassium wasting (20 meq in 24 h). Urinary excretion of uric acid, oxalate and phosphorus were in the normal range. Arterial blood gas analysis was normal with corresponding urine pH of 6.5. A furosemide and overnight fludrocortisone test (1 mg fludrocortisone 10 h pre-test/40 mg furosemide on day of test) was done for confirmation of incomplete distal renal tubular acidosis (RTA); urine pH remained at 6.5 confirming a distal acidification defect. There was no evidence of proximal tubular dysfunction. Plain radiograph and non-enhanced computed tomography (CT) of the abdomen showed left sided medullary nephrocalcinosis [Figure 2]. An intravenous pyelogram was done, which showed accumulation of contrast agent in the ectatic collecting tubules which persisted in delayed films and linear striations of contrast material radiating outward from the calyces (paint brush like appearance), [Figure 3].

Discussion

MSK is a developmental disorder with an estimated prevalence of 5/10,000 and 5/100,000.[1] It characterized by ectatic and cystic dilatations of the inner medullary collecting duct. It is usually considered as a sporadic disorder, but autosomal dominant pattern of inheritance has also been documented.[3] MSK remains clinically silent unless complicated by recurrent urinary tract infections or nephrolithiasis, most often it is a chance diagnosis on excretory urogram done for some other purpose. The disease can be segmental, unilateral or bilateral. Majority of MSK will have defects of urinary acidification without overt acid base disturbances.[4] The acidification defects are generally attributed secondary to the abnormal electrolyte transport resulting from the anatomic defects; but there are recent reports of occurrence of primary distal RTA in MSK as a result of H^+ ATPase mutations.[5] Excretory urography remains the corner stone of diagnosis. The classic appearance varies from papillary blush resulting from collections of contrast medium in ectatic papillary ducts in mild cases, linear striations and bouquets of flowers in full-fledged cases. Medullary nephrocalcinosis is seen in around 50% of cases and it is not mandatory for diagnosis. Ultrasound appearances are non-specific and may reveal a hyper echoic medulla. CT is less sensitive for picking up the characteristic findings except for medullary calcifications;[6] accuracy may be enhanced by 3D multi-detector-row CT urography.[7]

Congenital hemi hyperplasia has an estimated prevalence of 1 in 40,000, is associated with overgrowth of all somatic elements beyond the normal asymmetry.[8] Congenital hemi hyperplasia can be isolated (IHH) or part of well-defined hypertrophy syndromes such as Beckwith-Weidman Syndrome, Proteus syndrome, Russell-Silver syndrome, Neurofibromatosis type I (NF I) and Klippel-Trénaunay-Weber syndrome (KTW).[2,9] It may range from hyperplasia of the entire body or limited to a body part (segmental) or can be crossed. This may be associated with visceromegaly, other congenital anomalies like hypospadiasis and increased incidence of childhood embryonal neoplasms involving kidney and liver and adrenal carcinomas.[10] Isolated (non-syndromic) hemi hyperplasia occurs sporadically, but familial cases also has been reported. The association between MSK and hemi hyperplasia was first described by Steyn and

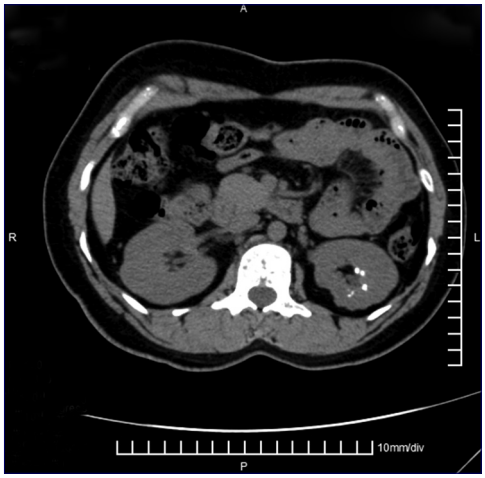

Figure 2: Unenhanced computed tomography showing left sided medullary nephrocalcinosis; note the right sided hyperplasia of the trunk as well as larger right kidney

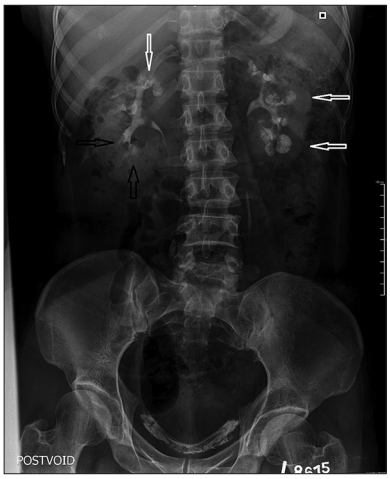

Figure 3: Intra venous pyelogram: (Delayed films) White arrowheads showing accumulation of contrast agent in the ectatic renal collecting tubule in the pyramids and linear striations of contrast material radiating outward from the calyces (paintbrushlike appearance); black arrow heads showing the papillary blush

Logic in 1964. Actual prevalence of MSK with IHH is not known, rough estimates are around 5-10%.[10] The MSK need not be limited to the side of hemihyperplasia, usually it is bilateral, can be unilateral or rarely contralateral.[8] There are no consensus criteria for the diagnosis of IHH; it can be so subtle that it may pass unnoticed as exemplified by the fact that in more than 50% of individuals with tumors in association with IHH, the diagnosis of IHH was made at the tumor diagnosis or later.[11]

As our patient does not have any childhood tumors or any other malformations associated with any of the known hypertrophy syndromes she is having IHH with MSK. To the best of our knowledge, this is the second case of IHH and MSK reported from India.

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