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Title: Bilaterally ectopic pelvic kidneys masquerading as horseshoe kidney in Fanconi anemia

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Abbreviated Title: Bilateral pelvic ectopic kidneys in FA

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Abstract:

Fanconi anemia (FA) is a genetic disease associated with the risk of different congenital malformations, bone marrow failure, and predisposition to cancer. Congenital abnormalities of the kidney and urinary tract are not infrequent in FA with renal ectopia being one of them. The incidence of the bilateral pelvic ectopic kidney is restricted to only a few reported cases; however, its association with FA has never been reported in the literature. We present a case of Fanconi anemia in a young girl with apparently fused kidneys on a ^{99m}Tc-DMSA planar scan which was confirmed to be bilateral pelvic kidneys on hybrid cross-sectional imaging.

Keywords: Fanconi anemia; bilateral pelvic kidney; DMSA; fused kidney; SPECT/CT.

Introduction:

Fanconi anemia (FA) is an inherited disease associated with various congenital and developmental abnormalities. The incidence of congenital abnormalities of the kidneys and the urinary tract is reported to be around 30% (*1*). It is important to include anatomical and functional imaging studies in the diagnostic workup of FA. Furthermore, renal disease is important for the management of FA at diagnosis as well as planning during the clinical course (*2*). To the best of our knowledge, the incidence of bilateral pelvic ectopic kidneys associated with FA has never been reported in the literature. We present an interesting case of FA with bilateral ectopic pelvic kidney masquerading as fused kidneys on ^{99m}Tc-DMSA planar scan which was confirmed to be bilateral pelvic kidneys on SPECT/CT imaging.

Case history:

An 11-year-old girl child presented to the pediatric emergency ward with complaints of easy fatiguability and decreased oral intake for the last 10 days. Her blood investigations revealed

pancytopenia. She received two packed red blood cells transfusions and underwent bone marrow aspiration to evaluate the cause of pancytopenia. She had similar complaints in the past for which she was given blood transfusions outside. Clinical manifestations and detailed laboratory tests revealed that the patient had FA. An x-ray abdomen followed by an ultrasound abdomen raised the possibility of fused and low-lying bilateral kidneys. She was referred to our department for functional imaging in the form of ^{99m}Tc-dimercaptosuccinic acid (DMSA) scan for the diagnosis of fused kidneys and functioning of its moieties. DMSA planar scan of the abdomen was taken 3 hours after intravenous injection of 92.5 MBq (2.5 mCi) of ^{99m}Tc DMSA. The images acquired in anterior, posterior, right, and left lateral views (Fig.1 A-B, E-F) revealed bilateral low-lying kidneys in the paramedian location fused in the midline. The differential functions for left and right moiety were 43% and 57% respectively. However, single positron emission computed tomography with low dose computed tomography (SPECT/CT) acquired for confirmation, revealed bilateral low-lying pelvic kidneys in the paramedian location that were separate from each other on trans-axial and coronal CT (Fig.1 C, G, arrows) sections with corresponding fused SPECT/CT images (Fig.1 D, H arrows).

Discussion:

FA is an inherited disease resulting from defects in FA/BRCA pathway for DNA interstrand crosslink repair. Around one-third to half of these patients may have associated congenital abnormalities of kidneys and urinary tract pointing to the deranged normal ascent of embryonic kidneys resulting in iliac/pelvic renal ectopia (*2,3*). The incidence of the ectopic pelvic kidney is around 1:2200-3000, however bilateral pelvic kidneys are a very rare developmental renal anomaly and are mostly diagnosed when patients develop symptoms due to obstruction, infection, and renal calculi (*4*). Anatomical and functional evaluation of renal malformations in

patients of FA is important for diagnostic work-up as well as for long-term management for achieving improved outcome with appropriate treatment. Every patient with renal abnormalities should be evaluated first with ultrasound followed by other imaging modalities. ^{99m}Tc-DMSA SPECT CT provides the advantage of both anatomical and functional evaluation for ectopic renal tissue anywhere from the thorax to the pelvis with a high degree of sensitivity in a single setting (*5*). Our case highlights the role of SPECT/CT to detect morphologic anomalies, cortical defects and avoid misinterpretation of findings on planar scintigraphy.

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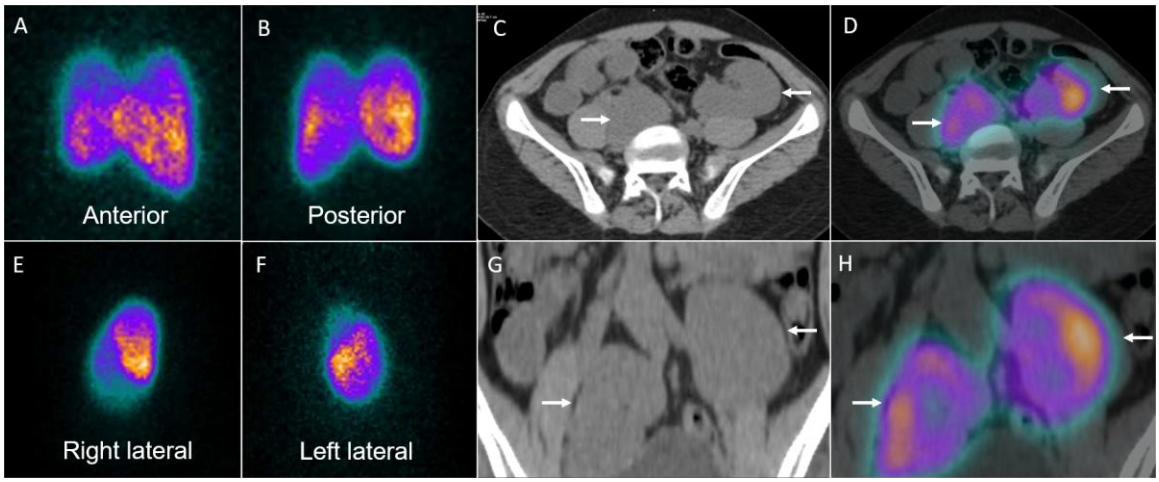

Fig 1: DMSA planar scan in anterior, posterior, right, and left lateral views (A-B, E-F) revealed bilateral low-lying kidneys in the paramedian location with both the kidneys to be apparently fused in the midline. Single photon emission computed tomography with low-dose computed tomography (SPECT/CT) was acquired which revealed bilateral low-lying pelvic kidneys in the paramedian location that was clearly separate from each other on trans-axial and coronal CT (C, G, arrows) sections with corresponding fused SPECT/CT images (D, H, arrows).