A female patient was found to have a horseshoe kidney on imaging studies when undergoing evaluation for recurrent urinary tract infections when she was in her 20s. Now in her late 40s, she presented with gross hematuria. A computed tomography urogram revealed the presence of a 14.9-cm mass arising from the isthmus of the horseshoe kidney (Figure 1). Also present was an enlarged retroperitoneal lymph node suspicious for metastatic disease.

Of the following tumor types, which has been described to occur with the highest frequency in individuals with a horseshoe kidney?

a. Wilms tumor  
b. Renal oncocytoma  
c. Urothelial carcinoma  
d. Primary well-differentiated neuroendocrine tumor

(see page 1688 for answer)
Answer: d. Primary well-differentiated neuroendocrine tumor.

Patients with horseshoe kidneys have been reported to have a two- to four-fold higher relative risk of Wilms tumor and urothelial carcinoma compared with the general population. However, the relative risk of primary well-differentiated neuroendocrine tumors (commonly referred to as carcinoid tumors) is significantly higher: estimated up to 62- to 82-fold higher in some studies. Furthermore, in studies of renal carcinoid tumors, approximately 17.8% to 19% of patients had horseshoe kidneys. It has been hypothesized that the “abnormal migration of posterior nephrogenic cells, which coalesce to form the isthmus of horseshoe kidneys” serves as the site of origin for these tumors.

This patient underwent a partial nephrectomy. The tumor cells were organized in tightly packed trabeculae with focal areas exhibiting nested and sheet-like growth (Figure 2A). The tumor cells exhibited a granular pattern of chromatin and lacked prominent nucleoli (Figure 2B). These morphologic features are consistent with a primary well-differentiated neuroendocrine tumor (carcinoid). In addition, a pre-aortic and inter-aortocaval lymph node dissection revealed metastatic disease in lymph nodes.

In summary, individuals with horseshoe kidneys have a higher risk of developing a primary well-differentiated neuroendocrine tumor (carcinoid). These patients frequently have involvement of regional lymph nodes at presentation and often have a prolonged clinical course despite the presence of metastatic disease.

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