Clinical Radiology Extra (2003) **58**: 35–37
doi:10.1016/S1477-6804(03)00005-0, available online at www.sciencedirect.com

Case Report

Renal Cell Carcinoma and Co-existing Polysplenia Syndrome

A. F. SCARSBROOK, D. R. WARAKAULLE, K. LAKIN

Department of Radiology, Northampton General Hospital, Cliftonville, Northampton, UK

INTRODUCTION

Polysplenia syndrome is an extremely rare constellation of congenital anomalies, with the main features being duplication of left-sided visceral organs and absent right-sided organs. We were able to find only two previous case reports of polysplenia associated with renal cell carcinoma. We report a third case of polysplenia syndrome with co-existing renal cell carcinoma where, as in both previous cases, the tumour arose from the left kidney. The imaging findings of polysplenia syndrome are presented, along with a brief review of the literature.

CASE REPORT

A 79 year-old woman presented to the Medical Outpatient Department with a history of progressive dyspnoea, wheeze and intermittent sputum production. Her lung function tests showed no significant abnormality, and she was referred to the Ear Nose and Throat clinic for evaluation of her pharynx. No pharyngeal abnormality was found on examination, and computed tomography (CT) was performed to evaluate her mediastinum. This showed termination of the IVC at the level of the right renal vein with azygos continuation (Figs 1*a*, 1*b* and 2). Both lungs showed left-sided morphology, with bilateral hyparterial bronchi (Fig. 1*b*) and an absent horizontal fissure on the right but were otherwise normal. There were two splenunculi (Fig. 2*b*) adjacent to a small spleen. The liver was located on the right side of the abdomen, and had a Riedel's lobe. A heterogeneously enhancing 4.5 cm mass was seen arising from the left kidney (Fig. 3). There was no evidence of lymphadenopathy, local invasion (apart from streaking of the perinephric fat) or distant metastases.

A left radical nephrectomy was performed. The spleen was found to be partly retroperitoneal at surgery, and the presence of two splenunculi was confirmed. Histology of the specimen confirmed it to be renal cell carcinoma. The cause for the patients initial symptoms was never elucidated but these settled after surgery.

Guarantor and correspondent: K. Lakin. Author for correspondence: Andrew F. Scarsbrook, Department of Radiology, John Radcliffe Hospital, Headley Way, Oxford OX3 9DU, UK. Tel: +44-1865-220825; Fax: +44-1865-220801; E-mail: andyscarsbrook1@aol.com

DISCUSSION

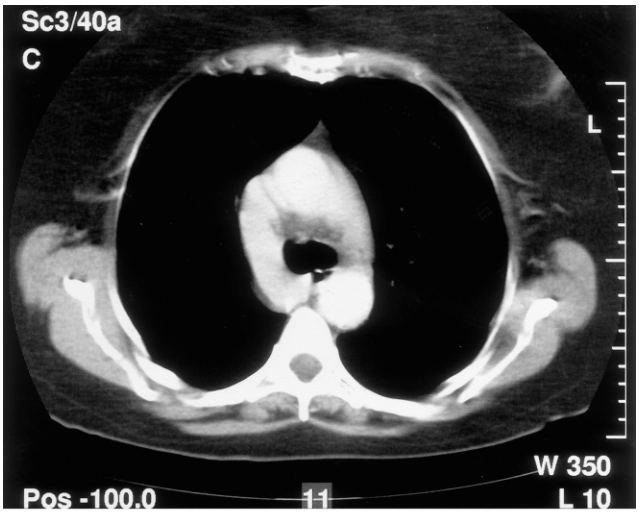
Polysplenia syndrome is part of the spectrum of visceral heterotaxias or situs ambiguous, which separate situs solitus (the normal arrangement of internal organs) from situs inversus, which is the complete reverse isomerism of the thoracic and abdominal viscera. Heterotaxias have been loosely subdivided into asplenia and polysplenia [1]. The polysplenia syndrome has a variable presentation, and its constituent features include:

(1) Congenital heart disease: anomalous pulmonary venous drainage, dextrocardia, atrial septal defect, endocardial cushion defect, pulmonary stenosis, transposition of the great arteries and double outlet right ventricle
(2) Gastrointestinal abnormalities: oesophageal atresia, tracheo–oesophageal fistula, gastric duplication, preduodenal portal vein, duodenal webs and atresia, short bowel, mobile caecum, malrotation, semi-annular pancreas, biliary atresia and absent gallbladder
(3) Genitourinary abnormalities: renal agenesis, renal and ovarian cysts
(4) Morphological abnormalities: bilateral left-sided, or rarely right-sided, lungs, bilateral superior vena cava (SVC), large azygos vein, multiple spleens or splenunculi, hepatic symmetry, and interrupted inferior vena cava (IVC) with azygos or hemiazygos continuation.

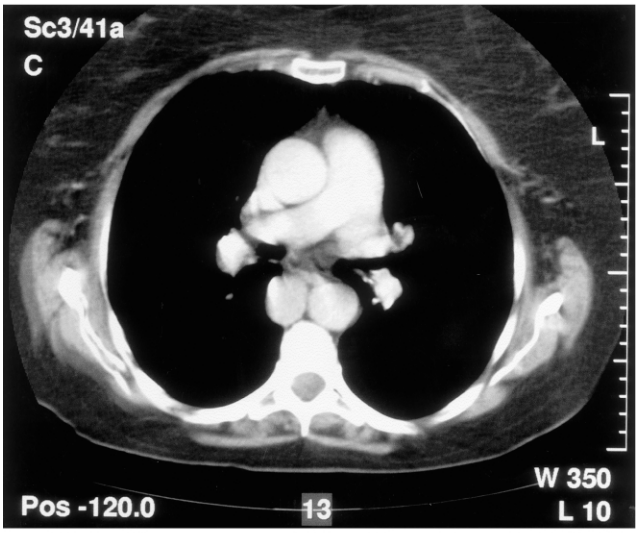
The most specific and consistent finding is a large azygos vein and arch, with associated IVC interruption [2].

Hirohata *et al*. [3] have previously reported two cases of renal cell carcinoma associated with polysplenia syndrome. The incidence of polysplenia syndrome is not known but it is thought to be very rare. Most patients with polysplenia syndrome die before the age of 1 year, with only a few surviving into adulthood [4]. It is theoretically possible, as in this case, that patients with less severe cardiac manifestations of the syndrome could evade diagnosis and hence the true incidence could be under estimated. Conversely the incidence of renal cell carcinoma is much more common occurring in one

1477-6804/03/$30.00/0

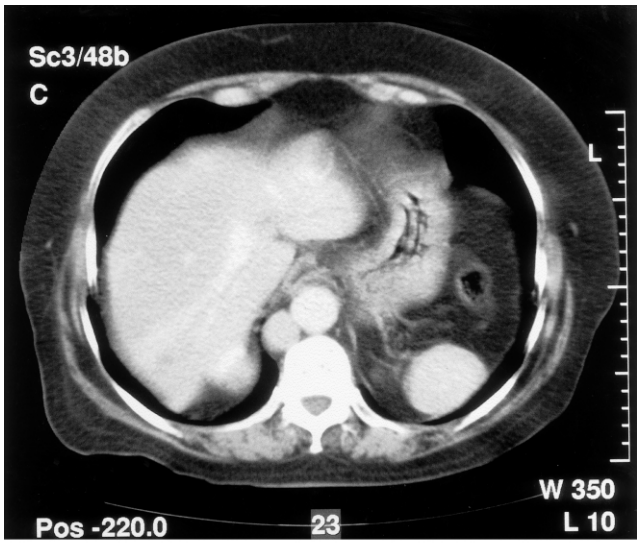


(*a*)

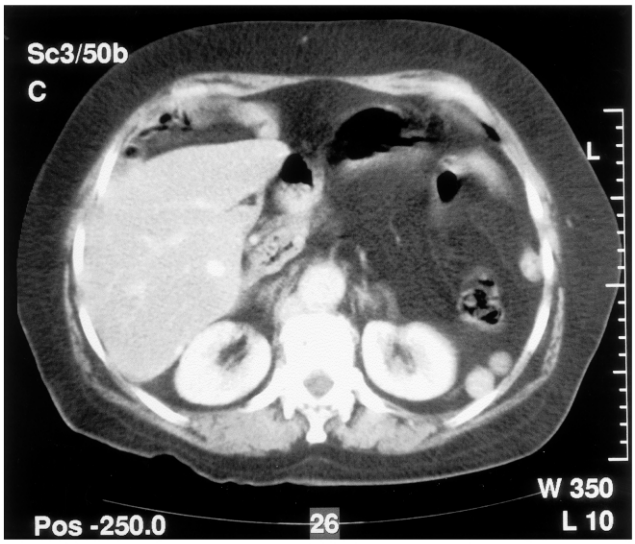


(*b*)

Fig. 1 – Axial CT sections through the thorax post-intravenous contrast medium administration. A dilated azygos arch (*a*) is seen, causing minor indentation of the carina. The intrathoracic azygos vein is also dilated, and there are bilateral hyparterial bronchi (*b*).



(*a*)



(*b*)

Fig. 2 – Axial CT sections through the upper abdomen post-intravenous contrast medium administration. The intrahepatic IVC is absent, and the spleen is small (*a*). There are two small left-sided splenunculi (*b*).

to three patients per 1000 [5,6]. It is the commonest renal tumour accounting for 85% of cases. Typically it occurs spontaneously in the fifth to seventh decade [7]. Even allowing for this it seems unlikely that the two are associated by chance alone.

Renal cell carcinoma is associated with other systemic anomalies occurring in up to 35% of patients with von Hippel-Lindau disease [8]. It has been reported in three cases of situs inversus totalis [9–11]. Polysplenia syndrome has also been reported in association with synchronous double cancers of the stomach and rectum [12]. In all of these case reports the associated tumour has been on the left side of the patient. It is important to note that all of the patients presumably had less severe forms of polysplenia syndrome without major cardiac involvement thus enabling them to survive into adulthood. It is most likely that the development of these tumours is coincidental given their relatively common incidence in adulthood. The majority of patients with renal cell carcinoma undergo staging CT of the chest and abdomen before surgical treatment and presumably some of the abnormalities associated with polysplenia syndrome should be visible on CT, as in our case. Given this, it is unlikely that there is a pathogenetic link between the two conditions since there have only been three documentated cases of the two co-existing in the world literature [3].

As in the case we have illustrated it may be that there are many more patients with undiagnosed polysplenia syndrome surviving into adulthood. We suggest that there is an absence of proof to support the theory that there is a common pathological link between heterotaxia and subsequent development of renal

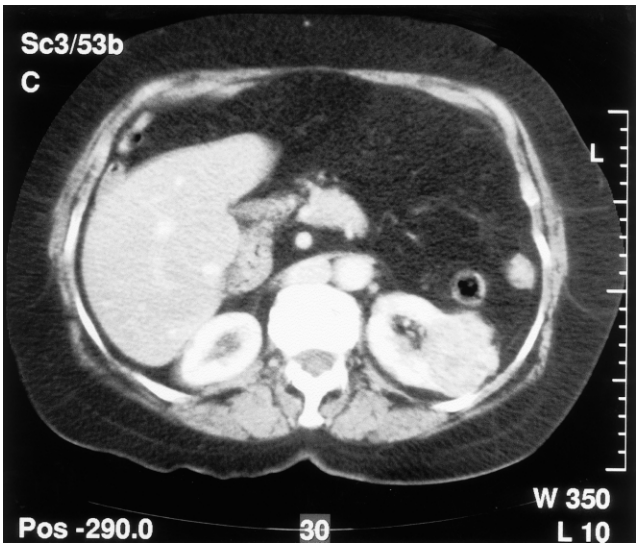


Fig. 3 – Axial CT post-intravenous contrast medium administration demonstrating the tumour arising from the left kidney.

or other abdominal malignancy, and that these cases are simply interesting observations rather than true associations. Nethertheless it is important to recognize the features of polysplenia syndrome given that it may in fact be more common than previously thought, and in order that its features are not mistaken for a pathological process.

REFERENCES

1 Ivermark BI. Implications of agenesis of the spleen on the pathogenesis of cono-truncus anomalies in childhood: analysis of the heart malformations in the splenic agenesis syndrome, with 14 new cases. *Acta Paediatr Scand*, 1955;44:1–110.
2 Dahnert W. Cardiovascular disorders. In: *Radiology Review Manual*, 4th edn Philadelphia: Lippincott Williams & Wilkins, 2000; 533–534.
3 Hirohata S, Isobe H, Mitamura T *et al.* Situs ambiguous with polysplenia complicated by renal adenocarcinoma. *Arch Intern Med*, 1985;145:1134–1137.
4 Rose V, Izukawa T, Moes CAF. Syndromes of asplenia and polysplenia: a review of cardiac and non-cardiac malformations in 60 cases with special reference to diagnosis and prognosis. *Br Heart J*, 1975;37:840–852.
5 Glenn JF. Renal tumours, In: Harrison JH, Gitter RF, Perlmutter AD, *et al. Campbell's Urology*. Philadelphia: W B Saunders Co, 1979, 967–1009.
6 Lakey WH. Tumors of the kidney, In: Karafin L, Kendall AR, eds. *Urology*. Hagerstown, MD: Harper & Row Publishers Inc, 1972;327–345.
7 Rottenberg G, Rankin S. Renal masses, In: Grainger RG, Allison DJ, Adam A, Dixon AK, eds. *Diagnostic Radiology*. London: Churchill Livingstone, 2001, 1557–1575.
8 Choyke PL, Glenn GM, Walther MM *et al.* von Hippel Lindau disease: genetic, clinical and imaging features. *Radiology*, 1995;194:629–642.
9 Treiger BF, Khazan R, Goldman SM *et al.* Renal cell carcinoma with situs inversus totalis. *Urology*, 1993;41:455–457.
10 Adler HL, Lerner SP. Renal cell carcinoma and situs inversus vicerum. *J Urol*, 1998;160:2141–2142.
11 Jimenez Verdejo A, Martinez Torres JL, Palao Yago F *et al.* Renal cell carcinoma in a patient with situs inversus and Kartagener syndrome. *Actas Urol Esp*, 2000;24:169–172.
12 Iwamura T, Shibata N, Haraguchi Y *et al.* Synchronous double cancer of the stomach and rectum with situs inversus totalis and polysplenia syndrome. *J Clin Gastroenterol*, 2001;33:148–153.