Case 12193

Imaging findings of renal cell carcinoma associated with Xp11.2 Translocation/TFE3 Gene Fusion in a young adult

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Section: Uroradiology & genital male imaging
Area of Interest: Kidney
Procedure: Diagnostic procedure
Technique: CT
Technique: MR
Special Focus: Neoplasia Case Type: Clinical Cases
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Patient: 31 years, male

Clinical History:

A 31-year-old man presented with a 1-month history of painless gross haematuria. No significant medical history was reported. A non-cystic mass lesion in the right kidney was found on ultrasound. Further imaging work-up with CT and MRI followed.

Imaging Findings:

CT disclosed the presence of a well-demarcated, expansile right renal mass lesion, measuring 7x4.5 cm in dimensions (Fig. 1-3). The mass was predominantly isodense to normal renal parenchyma in the precontrast scan and enhanced strongly and heterogeneously. Neither signs of perinephric fat invasion, nor retroperitoneal adenopathy were detected. The right renal vein and the inferior vena cava were patent. Based on CT findings, the diagnosis of renal malignancy, probably confined within the kidney was strongly suggested.

MRI followed revealing the presence of a thin hypointense halo around the tumour on T2-weighted sequences (Fig. 4), which proved to correspond to fibrous pseudocapsule on histology, confirming an early-stage disease. The solid parts of the renal mass showed restricted diffusion (Fig. 5) and strong, heterogeneous enhancement after gadolinium administration. The patient underwent right radical nephrectomy and pathology reported renal cell carcinoma associated with Xp11.2 translocation/TFE3 gene fusion (Xp11-RCC) (grade 3, stage pT2).

Discussion:

Renal cell carcinoma (RCC) associated with Xp11.2 translocation and transcription factor E3 (TFE3) gene fusion accounts for approximately 5-20% of RCCs in paediatric and adolescent patients [1-8]. These tumours are defined by several different translocations in chromosome Xp11.2 resulting in gene fusions in the TFE3 gene [1-8]. Macroscopically, Xp11.2 translocation RCCs usually display tan/yellow colour and are often accompanied by areas of necrosis and haemorrhage [1-8]. The tumour cells are polygonal, usually forming a papillary structure, with voluminous eosinophilic cytoplasm containing a few hyaline nodules and psammomatous calcifications [9]. The most distinctive immunohistochemical feature of this type of tumour is nuclear immunoreactivity for TFE3 protein [9]. The final diagnosis is based on microscopic appearance, TFE3 immunostaining and genetic analyses [1-8].

Xp11.2 RCC is typically presented as an asymptomatic renal mass often identified incidentally on imaging.
evaluation. Common clinical symptoms are gross haematuria, flank pain, or a palpable mass [7]. Prior exposure to chemotherapy is the only known risk factor.

Cross-sectional imaging, including CT and MRI is valuable for the diagnostic work-up and staging of this tumour. Typical Xp11.2 RCC usually presents with advanced stage at diagnosis, well-defined margins, cortical epicenter location, prevalence of intratumoural haemorrhage and/or calcifications, hypointensity on T2-weighted images, hypovascularity, with prolonged enhancement after contrast material administration [1-8]. Lesion hyperdensity on unenhanced CT images has also been reported, due to the presence of haemorrhage, proteinaceous fluid, numerous cellular components or combination of the above. Although, imaging findings are not pathognomonic, they should raise the suspicion for the presence of Xp11.2 RCC when detected in a child or young adult [5].

Radical nephrectomy is the primary treatment for Xp11.2 RCC. Like other RCCs, this tumour is not sensitive to radiotherapy or chemotherapy [1-8]. Because Xp11.2 translocation RCC has only recently been identified as an independent subtype of RCC, only few large cases series exist in the literature. Thus, little is known regarding its biological behaviour. At present, Xp11.2 translocation RCC is associated with poor prognosis, more often diagnosed at advanced stage, although this was not the case in our patient.

The liver, lungs and retroperitoneal lymph nodes are the most common metastatic sites [8].

**Differential Diagnosis List:** Renal cell carcinoma associated with Xp11.2 translocation/TFE3 gene fusion (Xp11-RCC), Papillary renal cell carcinoma, Clear cell renal cell carcinoma, Chromophobe renal cell carcinoma, Collecting duct renal cell carcinoma, Medullary carcinoma, Angiomyolipoma with minimal fat, Oncocytoma

**Final Diagnosis:** Renal cell carcinoma associated with Xp11.2 translocation/TFE3 gene fusion (Xp11-RCC)

**References:**


Description: Unenhanced axial CT image demonstrates a heterogeneous right renal mass (arrow) with central hypodense foci, corresponding to cystic change on pathology. Areas of calcifications were detected within the lesion. Origin: Department of Radiology University Hospital of Ioannina
Figure 2

Description: Coronal reformatted contrast-enhanced CT image (portal phase) shows a well-delineated right renal mass (arrow), heterogeneously and avidly enhancing. The tumour is located mainly in the interpolar region. Origin: Department of Radiology University Hospital of Ioannina
Description: Coronal reformatted contrast-enhanced CT image (nephrographic-excretory phase) depicts close proximity of the tumour (arrow) to the pelvicalyceal system, without obvious signs of invasion. Origin: Department of Radiology University Hospital of Ioannina
**Figure 4**

Description: Coronal T2-weighted image discloses an expansile right renal mass (arrow). The lesion is surrounded by a thin hypointense halo, due to the presence of incomplete fibrous pseudocapsule on pathology. **Origin:** Department of Radiology University Hospital of Ioannina
Description: Transverse ADC map demonstrates areas of restricted diffusion within the tumour (arrow).

Origin: Department of Radiology University Hospital of Ioannina