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Images in Clinical Medicine

Xp11 translocation renal cell carcinoma with vertebral metastasis presenting with low back pain and sciatica

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A 41-year-old male presented with lower back pain with the right posterior thigh and calf pain, gross hematuria, and accompanied by the bodyweight loss from 80 to 66 kg in recent months. The patient visited the local clinic and sciatica was impressed. Medication and rehabilitation were in vain; therefore, he visited our outpatient department of orthopedics. Magnetic resonance imaging (MRI) of the lumbar spine revealed bulky soft tissue replaced near the whole fifth lumbar vertebra, more at left aspect with destructive pattern and pathological fracture [Figure 1], and epidural mass with severe thecal sac compromised [Figure 2]. An exophytic tumor from the lower pole of the left kidney was found incidentally [Figure 1]. Further, the whole abdomen computed tomography (CT) demonstrated the same findings, also found some retroperitoneal lymphadenopathy (not shown). CT-guided biopsies were performed for the renal and lumbar vertebral lesions, respectively. The pathology showed solid-papillary neoplasms composed of epithelioid clear cells with strong nuclear labeling for transcription factor for immunoglobulin heavy-chain enhancer 3 (TFE3) proteins in both left renal [Figure 3] and vertebral [Figure 4] tumors, characteristic of Xp11 translocation renal cell carcinoma (RCC) with vertebral metastasis. Now, the patient is treated with target therapy, sunitinib, and radiotherapy.

Xp11 translocation RCC is a member of microphthalmia-associated transcription factor family translocation RCC, harboring gene fusions involving TFE3, which maps to Xp11,

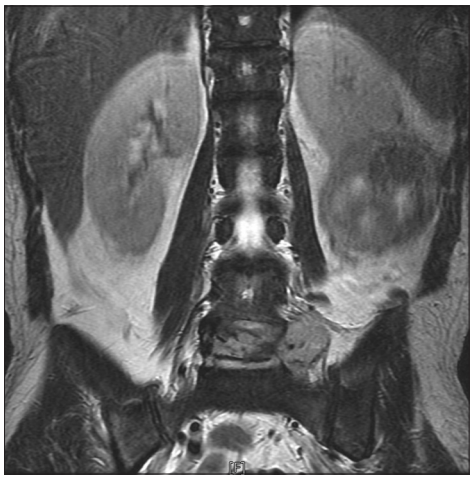

Figure 1: Magnetic resonance imaging of the lumbar spine revealed bulky soft tissue replaced near the whole fifth lumbar vertebra, more at left aspect with destructive pattern and pathological fracture, and an incidentally found exophytic tumor from the lower pole of the left kidney

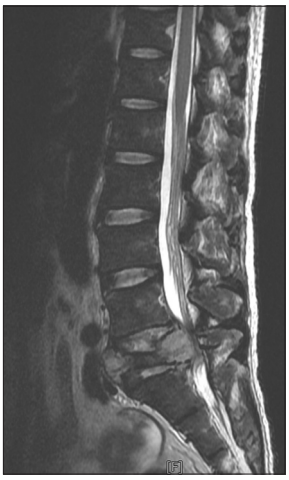

Figure 2: Magnetic resonance imaging of the lumbar spine revealed bulky soft tissue replaced near the whole fifth lumbar vertebra, with destructive pattern and pathological fracture, and epidural mass with severe thecal sac compromised

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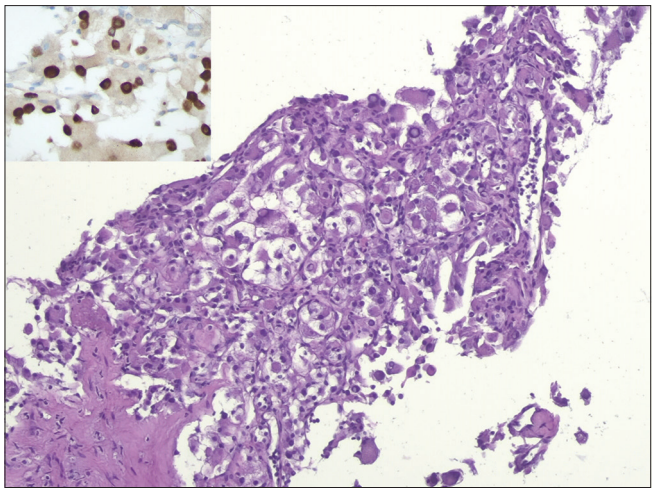

Figure 3: The pathology of the left renal tumor biopsy: Solid-papillary neoplasm composed of epithelioid clear cells (H and E, ×200) with strong nuclear labeling for transcription factor for immunoglobulin heavy-chain enhancer 3 proteins (inset, IHC, ×400), characteristic of Xp11 translocation renal cell carcinoma

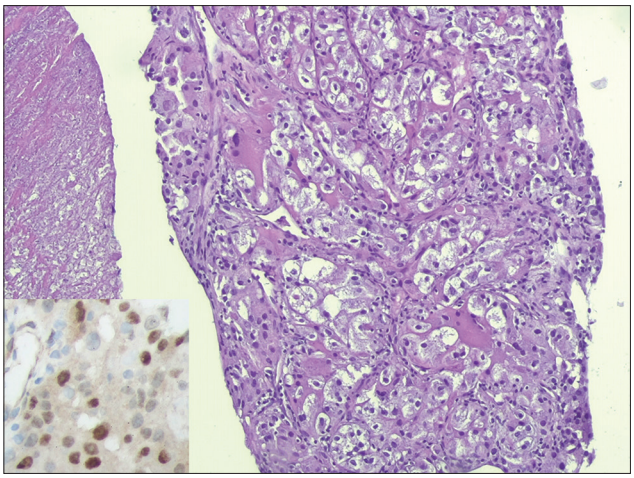

Figure 4: The pathology of vertebral tumor biopsy: Similar solid-papillary neoplasm composed of epithelioid clear cells with tumor necrosis (left field) (H and E, ×200) with strong nuclear labeling for transcription factor for immunoglobulin heavy-chain enhancer 3 proteins (inset, IHC, ×400), consistent with metastasis of Xp11 translocation renal cell carcinoma from the left renal tumor

with one of the multiple reported genes [1]. The two most common translocation of Xp11 translocation RCCs are t(X;1)(p11.2;q21), which fuses the papillary RCC and TFE3 genes, and t(X;17)(p11.2;q25), which fuses the alveolar soft part sarcoma chromosome region, candidate 1 (ASPSCR 1) and TFE3 genes. The fusions result in overexpression of aberrant TFE3 fusion proteins that activate expression of multiple downstream targets for tumorigenesis [2].

The most distinctive histopathologic morphology of Xp11 translocation RCCs is nested solid and papillary neoplasm composed of epithelioid clear cells [1]. The definite diagnosis is based on the strong nuclear TFE3 immunohistochemical staining, which is highly sensitive and specific [3], or TFE3 break-apart FISH assays, which have proven to be more useful because less susceptible to fixation issues [4].

Xp11 translocation RCCs are rare, accounting for 1.6%–4.0% of adult RCCs, and the median age is 41 years (range 15–59 years) [5]. The prognosis for patients with Xp11 translocation RCCs is similar to the patients with the most common type RCC-clear cell RCC, with most neoplasms presented at a low stage (pT1 or pT2) [5,6] and seldom (14%) distant metastasis at presentation [5]. Only distant metastasis and older age at diagnosis independently predicted death based on the multivariate analyses [7]. CT or MRI is useful for the evaluation of metastasis. The optimal treatment for Xp11 translocation RCC remains to be determined [5].

Declaration of patient consent

The authors certify that an appropriate patient consent form has been obtained. In the form, the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understands that his name and initials will not be published and due efforts will be made to conceal his identity.

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Conflicts of interest

There are no conflicts of interest.

References

1. Argani P, Antonescu CR, Couturier J, Fournet JC, Sciot R, Debiec-Rychter M, et al. PRCC-TFE3 renal carcinomas: Morphologic, immunohistochemical, ultrastructural, and molecular analysis of an entity associated with the t(X; 1)(p11.2;q21). Am J Surg Pathol 2002;26:1553-66.
2. Argani P, Lui MY, Couturier J, Bouvier R, Fournet JC, Ladanyi M. A novel CLTC-TFE3 gene fusion in pediatric renal adenocarcinoma with t(X; 17)(p11.2;q23). Oncogene 2003;22:5374-8.
3. Argani P, Lal P, Hutchinson B, Lui MY, Reuter VE, Ladanyi M. Aberrant nuclear immunoreactivity for TFE3 in neoplasms with TFE3 gene fusions: A sensitive and specific immunohistochemical assay. Am J Surg Pathol 2003;27:750-61.
4. Green WM, Yonescu R, Morsberger L, Morris K, Netto GJ, Epstein JI, et al. Utilization of a TFE3 break-apart FISH assay in a renal tumor consultation service. Am J Surg Pathol 2013;37:1150-63.
5. Komai Y, Fujiwara M, Fujii Y, Mukai H, Yonese J, Kawakami S, et al. Adult Xp11 translocation renal cell carcinoma diagnosed by cytogenetics and immunohistochemistry. Clin Cancer Res 2009;15:1170-6.
6. Pflueger D, Sboner A, Storz M, Roth J, Compérat E, Bruder E, et al. Identification of molecular tumor markers in renal cell carcinomas with TFE3 protein expression by RNA sequencing. Neoplasia 2013;15:1231-40.
7. Sukov WR, Hodge JC, Lohse CM, Leibovich BC, Thompson RH, Pearce KE, et al. TFE3 rearrangements in adult renal cell carcinoma: Clinical and pathologic features with outcome in a large series of consecutively treated patients. Am J Surg Pathol 2012;36:663-70.