Renal cell carcinoma associated with Xp11.2 translocation/transcription factor E3 gene fusion: A case report and literature review

DENGHAO YANG[1], WEN TANG[1], MINGWEN LIU[1], ZHIFEI XIE[1], FEI SHI[2], ZEJU ZHAO[1], XIAORONG YANG[3] and TAO WU[1]

[1]Department of Urology, The Affiliated Hospital of Zunyi Medical University; [2]Department of Urology, People's Hospital of Honghuagang District; [3]Department of Pathology, The Affiliated Hospital of Zunyi Medical University, Zunyi, Guizhou 563000, P.R. China

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Abstract. Renal cell carcinoma (RCC) associated with Xp11.2 translocation/transcription factor E3 (TFE3) gene fusion is a rare subtype of RCC. A 31-year-old male patient was admitted to The Affiliated Hospital of Zunyi Medical University (Zunyi, China) with a solid mass in the left kidney during a routine health examination. After ruling out surgical contraindications, the patient underwent a laparoscopic left partial nephrectomy under general anesthesia. Postoperative pathology and fluorescence *in situ* hybridization (FISH) identified Xp11.2 translocation RCC. There was no tumor recurrence or metastasis during the 1-year follow-up. Xp11.2 translocation RCC is unusual, its clinical and imaging findings are not specific, and the diagnosis depends on TFE3-immunohistochemical assay and FISH analysis. Surgical resection is the first choice of treatment and its prognosis is worse than that of clear cell RCC, thus regular follow-ups are necessary.

Introduction

Renal cell carcinoma (RCC) associated with Xp11.2 translocation/transcription factor E3 (TFE3) gene fusion is a rare subtype of RCC that was accepted as a distinct entity in the 2004 World Health Organization renal tumor classification (1). This type of RCC mainly occurs in young adults and children, accounting for 20-40% of cases of RCC; however, it is rarely seen in adults, with the proportion in adults only 1-1.6% global scale (2-4). The clinical manifestations of Xp11.2 translocation RCC are non-specific. Patients often seek medical attention due to symptoms such as hematuria, abdominal pain, or abdominal mass. However, asymptomatic patients who incidentally discover the tumors during physical examination. The present study describes the case of a patient with Xp11.2 translocation RCC, who was admitted to the Department of Urology (The Affiliated Hospital of Zunyi Medical University; Zunyi, China). No tumor recurrence or metastasis was found in the 1-year follow-up and the present study retrospectively analyzed the diagnosis and treatment of the patient, and reviewed the relevant literature, aiming to improve the understanding of the symptoms, diagnosis, treatment and prognosis of RCC.

Case report

A 31-year-old man was diagnosed with a solid mass in the left kidney during a routine health examination at the Department of Urology (The Affiliated Hospital of Zunyi Medical University) in October 2022. Upon tracing their medical history, the patient had reported experiencing lower abdominal pain and lower back pain for several months. However, they denied experiencing symptoms such as fever, stomachache, hematuria, frequent urination, urinary hesitancy and weight loss. The patient had no previous history of hypertension or diabetes, no surgical history, no family history of cancer and did not have a history of smoking or drinking alcohol. Before the operation, the patient did not present any significant abnormalities in their physical signs, such as coughing, fever, and respiratory distress. Routine blood routine tests, blood biochemistry tests, and a coagulation test, also did not detect any notable abnormalities.

An abdominal CT scan revealed a well-defined left renal tumor measuring 2.6x3.3x2.8 cm, slightly enhanced at the arterial phases. No evidence of metastases or abnormalities in the right kidney was found (Fig. 1A and B). Based on the clinical diagnosis, the left kidney tumor was considered to be clear cell RCC (ccRCC). After ruling out surgical contraindications, the patient underwent a laparoscopic left partial nephrectomy under general anesthesia 8 days after the routine health examination. The operation was successful, and the

Correspondence to: Dr Tao Wu, Department of Urology, The Affiliated Hospital of Zunyi Medical University, 145 Dalian Road, Huichuan, Zunyi, Guizhou 563000, P.R. China
E-mail: martin0600@163.com

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tumor was completely removed, measuring 3x2x2 cm in size and presenting as solid, with a cut surface that was tan-yellow, segmentally demonstrating hemorrhagic areas (Fig. 2A and B). After resection, the tumor was sent to the Department of Pathology (Affiliated Hospital of Zunyi Medical University) for H&E and immunohistochemical staining. Several common kidney tumor markers were assessed, including CD10, cytokeratin7 (CK7), vimentin, Succinate Dehydrogenase Iron-Sulfur Subunit, P504S, carbonic anhydrase 9 and TFE3. Ultimately, the four typical indicators, CD10, CK7, vimentin and TFE3, were selected to support the diagnosis of Xp11.2 translocation RCC. Microscopic examination indicated that the tumor cells had a papillary or nested architecture and consisted of cells with voluminous, clear or eosinophilic cytoplasm, and it was observed that these structures contained fibrovascular cores and the focal presence of psammoma bodies (Fig. 3A-D). Immunohistochemistry (IHC) showed clear and diffuse expression of TFE3 and CD10 (Fig. 3E and F), while vimentin and CK7 were negative (Fig. 3G and H). The results of the immunohistochemical staining supported the diagnosis of Xp11.2 translocation RCC.

To confirm the diagnosis, the pathologist recommended a fluorescence *in situ* hybridization (FISH) assay targeting the TFE3 gene. After obtaining the consent of the patient, the samples were sent to ShengTingGroup for the FISH assay. The FISH assay showing the split orange (centromeric side) and green (telomeric side) signals (white arrow) and quantitative analysis demonstrated that the number of cells isolated by orange and green signal was 80% (>15%) indicating TFE3 gene rearrangement (Fig. 4). Consequently, the patient was eventually diagnosed with Xp11.2 translocation RCC of the right kidney. Following the surgery, the patient had a smooth recovery and was discharged after 2 weeks. Subsequent follow-up examinations over a period of 6 months revealed no tumor recurrence or metastasis.

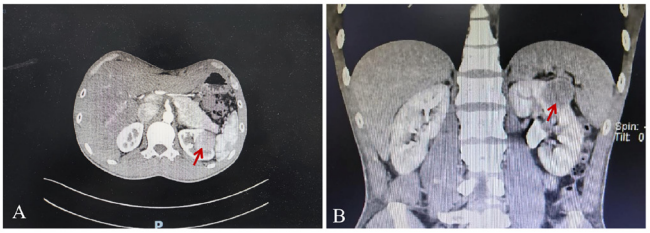

Figure 1. Results of the abdominal CT scan. (A) A well-circumscribed and slightly heterogeneous enhancing mass of the left kidney in the arterial phase (red arrow). (B) A 3D reconstruction showed that the tumor was a 2.6x3.3x2.8 cm mass with a solid component and located in the left renal (red arrow). The right kidney was normal.

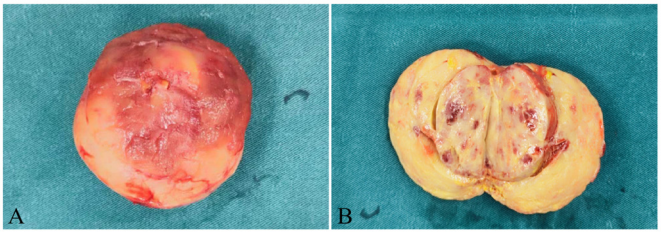

Figure 2. Appearance of the removed tumor. (A) Tumor was completely removed, measuring 3x2x2 cm in size. (B) Cross-section of the tumor presented as solid, with a cut surface that was tan-yellow, and segmentally demonstrated hemorrhagic areas.

Discussion

Xp11.2 translocation RCC with TFE3 gene fusion is a rare subtype of RCC (5). It occurs due to a gene fusion between the TFE3 gene on the Xp11.2 chromosome and one of six different fusion partners (6). This fusion leads to overexpression of the TFE3 protein, which in turn upregulates the MET tyrosine kinase receptor, activating downstream signaling pathways that promote cell proliferation and tumor formation (7). Xp11.2 translocation RCC differs from other types of renal carcinoma in terms of histology, immunophenotype and prognosis (8). As a result, it has been classified as a rare and unique subtype of RCC in the renal tumor classification of The World Health Organization from the 2004 edition onwards (9). The molecular identity of five out of the six known gene fusion partners of TFE3 are papillary renal cell Carcinoma, Alveolar Soft Part Sarcoma, polypyrimidine tract-binding protein-associated splicing factor, non-POU domain-containing octamer-binding and clathrin heavy-chain genes, while the identity of the sixth partner on chromosome 3 is still unknown (10-12).

The incidence of Xp11.2 translocation RCC is relatively low. Previous studies have reported its incidence as 0.9% of adult RCC cases, 15% of young adult RCC cases and 54% of child RCC cases (3,13-15). Although Xp11.2 translocation RCC is more common in children, the overall number of cases may be higher in adults due to the higher prevalence of RCC in that population (16). Additionally, sex differences in the incidence of Xp11.2 translocation RCC may exist, given that these translocations occur on the X chromosome (15,17). A meta-analysis revealed a higher incidence of Xp11.2 translocation RCC in women compared with in men, with an adult female-to-male ratio of 1.6-3.6:1. However, this sex difference is not evident in pediatric patients (16).

Xp11.2 translocation RCC shares similarities with other types of RCC and lacks specific clinical manifestations. Patients with Xp11.2 translocation RCC typically present to the hospital with symptoms such as hematuria, abdominal pain or an abdominal mass (known as the triad of RCC) (18). However, most patients only experience one of these symptoms. Gross hematuria is the most common initial symptom, while cases presenting with the complete triad of RCC are extremely rare (19). Some patients may have distant metastasis symptoms as their primary manifestation, including bone pain, dull pain in the liver area or hemoptysis (19). However, numerous patients with Xp11.2 translocation RCC are asymptomatic, and their tumors are incidentally discovered during physical examinations (20). The tumors are typically located in the right kidney in ~66.7% of cases and in the left kidney in ~3.3% of cases (21).

On CT and MRI scans, Xp11.2 translocation RCC is often indistinguishable from the more common ccRCC (22). However, there are still some characteristic features that can help differentiate between them. While both types can exhibit low, equal or high density on unenhanced CT scans, Xp11.2 translocation RCC generally demonstrates less enhancement compared with ccRCC on enhanced CT scans (22,23). This

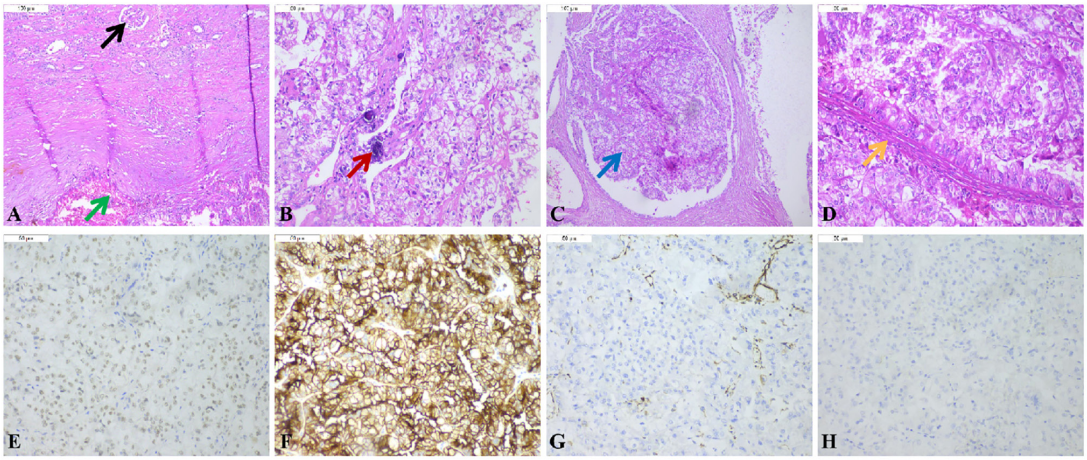

Figure 3. Pathological features of the mass. (A) Presence of normal kidney tissue and tumor tissue (black arrow indicates the glomerulus; green arrow indicates the tumor tissue; H&E staining; magnification, x100). (B) The tumor was composed of a single population of small, round to oval-shaped cells with voluminous, clear or eosinophilic cytoplasm. Psammoma bodies of the tumor were also observed (red arrow indicates the psammoma bodies; magnification, x200). (C) Tumor cells were arranged in nested architecture (blue arrow; magnification, x100). (D) Fibrovascular cores of papillary structures were present (orange arrow; magnification, x200). (E) Positive immunostaining of the tumor cells for TFE3 expression (magnification, x200). (F) Positive immunostaining of the tumor cells for CD10 expression (magnification, x200). (G) Negative immunostaining for vimentin in the tumor cells (magnification, x200). (H) Negative immunostaining for CK7 in the tumor cells (magnification, x200). CK7, cytokeratin 7; TFE3, transcription factor E3.

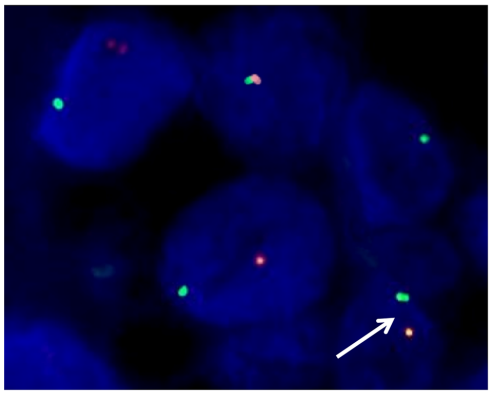

Figure 4. TFE3 fluorescence *in situ* hybridization assay showing the split orange and green signals (white arrow); the number of cells isolated by orange and green signal is 80% (>15%) indicating TFE3 gene rearrangement (magnification, x1,000). TFE3, transcription factor E3.

is attributed to the presence of fewer blood vessels within the focal area of Xp11.2 translocation RCC. Furthermore, on MRI scans, Xp11.2 translocation RCC typically exhibits slightly high signal intensity on T1-weighted images before angiography, and equal-to-low signal intensity on T2-weighted MR images (24). These findings are in contrast to the results observed in cases of ccRCC. Considering the age of the patient, if they are children or adolescents, a diagnosis of Xp11.2 translocation RCC should be highly considered. This distinction holds important guiding significance for the development of a preoperative treatment plan (25).

The morphological features of Xp11.2 translocation RCC often overlap with those of other types of RCC. Macroscopically, the tumor typically presents as solid and occasionally cystic, with a cut surface that is mostly gray or tan-yellow, occasionally demonstrating necrotic or hemorrhagic areas (23). These features are similar to what is observed in ccRCC (26). However, there are histological characteristics that can help distinguish Xp11.2 translocation RCC from other subtypes. The most distinct histological appearance of Xp11.2 translocation RCC is the presence of a papillary structure comprised of clear cells and eosinophilic cells. These structures contain fibrovascular cores and often exhibit psammoma bodies, which are rarely seen in conventional ccRCC (27,28). The frequency of psammoma bodies has been reported as 50 and 62% in two clinicopathological studies (29,30). Although the tumor may have a relatively typical histological appearance, an accurate diagnosis still requires supporting immunohistochemical findings. IHC typically shows minimal reactivity to CK7 or vimentin (31). Overexpression of the TFE3 protein is observed due to translocation, and studies have demonstrated the sensitivity (97.5%) and specificity (99.6%) of TFE3 protein expression in diagnosing Xp11.2 translocation RCC (5,31). Therefore, TFE3 protein expression detected by IHC is the most commonly utilized auxiliary diagnostic technique in clinical practice (32). However, immunohistochemical results can be influenced by factors such as immunostaining methods, incubation methods, fixation time, antigen repair methods, and antibody sensitivity and specificity, all of which can potentially lead to false-positive or false-negative results (33). Qu *et al* (33) reported a false-positive rate of 6.7% and a false-negative rate of 4.3% in the diagnosis of Xp11.2 translocation RCC using TFE3-IHC. Therefore, further tests are essential to validate the TFE3 immunohistochemical results in order to achieve a more accurate diagnosis (34). Common methods currently used for validation include karyotype analysis, reverse transcription polymerase chain reaction (RT-PCR) and FISH (34,35). Among these, karyotype analysis has the highest accuracy, but it requires fresh tissue samples and, clinically, tumor tissue excised during surgery is typically preserved and fixed in formaldehyde solution, rendering it unsuitable as a common diagnostic tool (36). However, formalin-fixed paraffin-embedded tissues can be used for FISH and/or RT-PCR analysis, although the latter method

is rarely employed for diagnosis due to RNA degradation in archived materials (37). FISH has emerged as a preferred method, offering cheapness, speed, convenience and accuracy, making it the gold standard for clinical diagnosis of Xp11.2 translocation RCC (38,39). Given the unclear pathological type prior to surgery, the treatment plan for Xp11.2 translocation RCC often follows that of ccRCC. For tumors measuring <4 cm, partial nephrectomy is recommended; for tumors >4 cm without metastasis, radical nephrectomy is advised (18,40); and targeted therapy is recommended for patients with distant metastasis (33). The overall prognosis for Xp11.2 translocation RCC is generally worse than that of ccRCC, with a more favorable prognosis observed in children compared with adults (41). Zhong *et al* (42) reported that children and adolescents with lymph node-positive Xp11.2 translocation RCC in the absence of distant metastases had a favorable prognosis, with overall survival estimates nearly triple those of adult patients with a similar presentation. Therefore, if postoperative pathology confirms Xp11.2 translocation RCC, more frequent follow-ups are recommended compared with that for ccRCC. Additionally, adults with confirmed Xp11.2 translocation RCC based on postoperative pathology should be followed up more frequently than children and adolescents.

In summary, Xp11.2 translocation RCC is relatively rare, and its clinical and imaging manifestations lack specificity. The combination of TFE3-IHC assay and FISH assay is an accurate and effective method for separately screening and confirming the diagnosis of Xp11.2 translocation RCC. Surgical resection without metastasis is the preferred method, and targeted therapy is recommended in cases of metastasis. Its prognosis is worse than that of ccRCC, and the prognosis of adults is significantly lower than that of children and adolescents, thus regular follow-ups are important.

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Availability of data and materials

The datasets used and/or analyzed during the current study are available from the corresponding author on reasonable request.

Authors' contributions

TW conceived and the design of the study. DY analyzed and interpretation of the data and revised the manuscript drafting critical revisions on the intellectual content. WT, ML, ZX and FS acquired and analyzed the data. ZZ and XY analyzed and interpreted the data. DY and TW confirm the authenticity of all the original data. All authors have reviewed and approved the final manuscript.

Ethics approval and consent to participate

Not applicable.

Patient consent for publication

Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

Competing interests

The authors declare that they have no competing interests.

References

1. Pipitone S, Vitale MG, Baldessari C, Dominici M and Sabbatini R: Long survival of a young patient with Xp11.2 translocation metastatic clear cell renal carcinoma: Case report. Tumori 107: NP131-NP135, 2021.
2. Kmetec A and Jeruc J: Xp 11.2 translocation renal carcinoma in young adults; recently classified distinct subtype. Radiol Oncol 48: 197-202, 2014.
3. Komai Y, Fujiwara M, Fujii Y, Mukai H, Yonese J, Kawakami S, Yamamoto S, Migita T, Ishikawa Y, Kurata M, *et al*: Adult Xp11 translocation renal cell carcinoma diagnosed by cytogenetics and immunohistochemistry. Clin Cancer Res 15: 1170-1176, 2009.
4. Zhu Y, Pu X, Dong X, Ji C, Guo H, Li D, Zhao X and Gan W: Molecular heterogeneity of Xp11.2 translocation renal cell carcinoma: The correlation between split signal pattern in fish and prognosis. Cancer Manag Res 13: 2419-2431, 2021.
5. Guo W, Zhu Y, Pu X, Guo H and Gan W: Clinical and pathological heterogeneity of four common fusion subtypes in Xp11.2 translocation renal cell carcinoma. Front Oncol 13: 1116648, 2023.
6. Karashima T, Kuno T, Kuroda N, Satake H, Fukata S, Chikazawa M, Kawada C, Yamasaki I, Shuin T, Hiroi M and Inoue K: Bilateral Xp11.2 translocation renal cell carcinoma: A case report. BMC Urol 18: 106, 2018.
7. Wang P, Zhang X, Shao SH, Wu F, Du FZ, Zhang JF, Zuo ZW and Jiang R: Chemotherapy, transarterial chemoembolization, and nephrectomy combined treated one giant renal cell carcinoma (T3aN1M1) associated with Xp11.2/TFE3: A case report. World J Clin Cases 10: 10180-10185, 2022.
8. Ellis CL, Eble JN, Subhawong AP, Martignoni G, Zhong M, Ladanyi M, Epstein JI, Netto GJ and Argani P: Clinical heterogeneity of Xp11 translocation renal cell carcinoma: Impact of fusion subtype, age, and stage. Mod Pathol 27: 875-886, 2014.
9. Lopez-Beltran A, Scarpelli M, Montironi R and Kirkali Z: 2004 WHO classification of the renal tumors of the adults. Eur Urol 49: 798-805, 2006.
10. Argani P, Lui MY, Couturier J, Bouvier R, Fournet JC and Ladanyi M: A novel CLTC-TFE3 gene fusion in pediatric renal adenocarcinoma with t(X;17)(p11.2;q23). Oncogene 22: 5374-5378, 2003.
11. Clark J, Lu YJ, Sidhar SK, Parker C, Gill S, Smedley D, Hamoudi R, Linehan WM, Shipley J and Cooper CS: Fusion of splicing factor genes PSF and NonO (p54nrb) to the TFE3 gene in papillary renal cell carcinoma. Oncogene 15: 2233-2239, 1997.
12. Mathur M, Das S and Samuels HH: PSF-TFE3 oncoprotein in papillary renal cell carcinoma inactivates TFE3 and p53 through cytoplasmic sequestration. Oncogene 22: 5031-5044, 2003.
13. Argani P and Ladanyi M: Translocation carcinomas of the kidney. Clin Lab Med 25: 363-378, 2005.
14. Ramphal R, Pappo A, Zielenska M, Grant R and Ngan BY: Pediatric renal cell carcinoma: Clinical, pathologic, and molecular abnormalities associated with the members of the mit transcription factor family. Am J Clin Pathol 126: 349-364, 2006.
15. Sukov WR, Hodge JC, Lohse CM, Leibovich BC, Thompson RH, Pearce KE, Wiktor AE and Cheville JC: 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 36: 663-670, 2012.
16. Xu L, Yang R, Gan W, Chen X, Qiu X, Fu K, Huang J, Zhu G and Guo H: Xp11.2 translocation renal cell carcinomas in young adults. BMC Urol 15: 57, 2015.

17. Wu Y, Chen S, Zhang M, Liu K, Jing J, Pan K, Zhang L, Xu B, Lu X and Chen M: Factors associated with survival from Xp11.2 translocation renal cell carcinoma diagnosis-A systematic review and pooled analysis. Pathol Oncol Res 27: 610360, 2021.
18. Wang Y, Wang Y, Feng M, Lian X, Lei Y and Zhou H: Renal cell carcinoma associated with Xp11.2 translocation/transcription factor E3 gene fusion: An adult case report and literature review. J Int Med Res 48: 300060520942095, 2020.
19. Hung CC, Pan CC, Lin CC, Lin ATL, Chen KK and Chang YH: XP11.2 translocation renal cell carcinoma: Clinical experience of Taipei Veterans General Hospital. J Chin Med Assoc 74: 500-504, 2011.
20. Armah HB and Parwani AV: Xp11.2 translocation renal cell carcinoma. Arch Pathol Lab Med 134: 124-129, 2010.
21. Lim B, You D, Jeong IG, Kwon T, Hong S, Song C, Cho YM, Hong B, Hong JH, Ahn H and Kim CS: Clinicopathological features of Xp11.2 translocation renal cell carcinoma. Korean J Urol 56: 212-217, 2015.
22. Chen X, Zhu Q, Li B, Cui W, Zhou H, Duan N, Liu Y, Kundra V and Wang Z: Renal cell carcinoma associated with Xp11.2 translocation/TFE gene fusion: Imaging findings in 21 patients. Eur Radiol 27: 543-552, 2017.
23. Dong H, Ni Y, Liu Z, Wang Z, Hu B, Xu H and Cai S: Imaging findings, clinical and pathological characters of 28 patients with Xp11.2/TFE3 translocation renal cell carcinoma. J Cancer Res Ther 19: 132-140, 2023.
24. Wang W, Ding J, Li Y, Wang C, Zhou L, Zhu H and Peng W: Magnetic resonance imaging and computed tomography characteristics of renal cell carcinoma associated with Xp11.2 translocation/TFE3 gene fusion. PLoS One 9: e99990, 2014.
25. Liu K, Xie P, Peng W and Zhou Z: Renal carcinomas associated with Xp11.2 translocations/TFE3 gene fusions: Findings on MRI and computed tomography imaging. J Magn Reson Imaging 40: 440-447, 2014.
26. Ge Y, Lin X, Zhang Q, Lin D, Luo L, Wang H and Li Z: Xp11.2 translocation renal cell carcinoma with TFE3 rearrangement: Distinct morphological features and prognosis with different fusion partners. Front Oncol 11: 784993, 2021.
27. Al-Maghrabi JA and Khabaz MN: Uncommon localization of extrarenal Xp11.2 translocation-associated renal cell carcinoma (RCC): Case report. Appl Immunohistochem Mol Morphol 28: e33-e35, 2020.
28. Zhou J, Zhao L, Yang Z, Chen Y, Wu X and Xue W: Clinicopathologic, treatment and prognosis study of 46 Xp11.2 translocation/TFE3 gene fusion renal cell carcinomas. BMC Urol 22: 109, 2022.
29. Liu N, Qu F, Wei K, Gan W, Wang Z, Zhuang W, Agizamhan S, Ma W, Yang J, Chen M, *et al*: Incidence and significance of psammoma bodies in Xp11.2 translocation renal cell carcinoma and papillary renal cell carcinoma. Oncol Lett 18: 472-478, 2019.
30. Manucha V, Sessums MT, Lewin J and Akhtar I: Cyto-histological correlation of Xp11.2 translocation/TFE3 gene fusion associated renal cell carcinoma: Report of a case with review of literature. Diagn Cytopathol 46: 267-270, 2018.
31. Argani P, Lal P, Hutchinson B, Lui MY, Reuter VE and Ladanyi M: Aberrant nuclear immunoreactivity for TFE3 in neoplasms with TFE3 gene fusions: A sensitive and specific immunohistochemical assay. Am J Surg Pathol 27: 750-761, 2003.
32. Dey B, Badhe B, Govindarajan KK and Ramesh RA: Xp11.2 translocation renal cell carcinoma diagnosed by immunohistochemistry and cytogenetics. J Lab Physicians 8: 123-125, 2016.
33. Qu Y, Gu C, Wang H, Chang K, Yang X, Zhou X, Dai B, Zhu Y, Shi G, Zhang H and Ye D: Diagnosis of adults Xp11.2 translocation renal cell carcinoma by immunohistochemistry and FISH assays: Clinicopathological data from ethnic Chinese population. Sci Rep 6: 21677, 2016.
34. Klatte T, Streubel B, Wrba F, Remzi M, Krammer B, de Martino M, Waldert M, Marberger M, Susani M and Haitel A: Renal cell carcinoma associated with transcription factor E3 expression and Xp11.2 translocation: Incidence, characteristics, and prognosis. Am J Clin Pathol 137: 761-768, 2012.
35. Gaillot-Durand L, Chevallier M, Colombel M, Couturier J, Pierron G, Scoazec JY and Mege-Lechevallier F: Diagnosis of Xp11 translocation renal cell carcinomas in adult patients under 50 years: Interest and pitfalls of automated immunohistochemical detection of TFE3 protein. Pathol Res Pract 209: 83-89, 2013.
36. Zhuang W, Liu N, Guo H, Zhang C and Gan W: Gender difference analysis of Xp11.2 translocation renal cell carcinomas's attack rate: A meta-analysis and systematic review. BMC Urol 20: 130, 2020.
37. Malouf GG, Su X, Yao H, Gao J, Xiong L, He Q, Compérat E, Couturier J, Molinié V, Escudier B, *et al*: Next-generation sequencing of translocation renal cell carcinoma reveals novel RNA splicing partners and frequent mutations of chromatin-remodeling genes. Clin Cancer Res 20: 4129-4140, 2014.
38. Kim SH, Choi Y, Jeong HY, Lee K, Chae JY and Moon KC: Usefulness of a break-apart FISH assay in the diagnosis of Xp11.2 translocation renal cell carcinoma. Virchows Arch 459: 299-306, 2011.
39. Rao Q, Williamson SR, Zhang S, Eble JN, Grignon DJ, Wang M, Zhou XJ, Huang W, Tan PH, Maclennan GT and Cheng L: TFE3 break-apart FISH has a higher sensitivity for Xp11.2 translocation-associated renal cell carcinoma compared with TFE3 or cathepsin K immunohistochemical staining alone: Expanding the morphologic spectrum. Am J Surg Pathol 37: 804-815, 2013.
40. Song HC, Sun N, Zhang WP, He L, Fu L and Huang C: Biological characteristics of pediatric renal cell carcinoma associated with Xp11.2 translocations/TFE3 gene fusions. J Pediatr Surg 49: 539-542, 2014.
41. Dong J, Xu W, Ji Z and Pan B: Xp11.2 translocation renal cell carcinoma: Clinical characteristics and potential prognostic predictors. Dis Markers 2021: 5647933, 2021.
42. Zhong M, De Angelo P, Osborne L, Paniz-Mondolfi AE, Geller M, Yang Y, Linehan WM, Merino MJ, Cordon-Cardo C and Cai D: Translocation renal cell carcinomas in adults: A single-institution experience. Am J Surg Pathol 36: 654-662, 2012.