Original Research Article

Rare case of renal tumor : Carcinoma with Xp11 translocation

Absract :

Adult renal cell carcinomas are divided into clear cell carcinomas (75%), papillary carcinomas (10%) and chromophobe carcinomas. Cytogenetic analysis has made it possible to discover new variants of renal cell carcinomas. In the 2004 WHO classification, a rare entity, "Xp11.2 translocation-related carcinoma", has appeared.

The patient was T.A., aged 21, with no particular pathological history. The history of disease goes back to 5 years with the appearance of a total intermittent clotting hematuria without any other associated sign.

The urogenital examination found a lumbar tenderness with a positive left lumbar contact

A biological check-up was requested showing an anemia of 6.5 g/dl, grouping: B+.

A normal renal function with a creatinemia of 11 mg /l

A radiological workup (Uroscanner) was also requested, showing a large left renal tumor process occupying almost the entire kidney, estimated at 19.5*14*12.5 cm in the major axes

The patient underwent a packed red blood cell transfusion and then an enlarged total nephrectomy with a pre aortic-cavity lymph node dissection.

The genetic examination is the reference diagnosis. It is requested in the first instance to allow a decision to be made after an equivocal immunohistochemical examination.

The evolution of tRCC is often negative in adults, with a high risk of lymph node metastases. The evolution of our patient was marked by a good clinical and biological improvement with absence of low back pain, hematuria and normalization of the hemoglobin level.

Keyword : tumor, kidney, Carcinoma with Xp11 translocation

Introduction :

The classification of renal cell carcinomas is based on morphologic criteria such as tumor cell architecture and cytology. Among adult renal cell carcinomas, we distinguish clear cell carcinomas (75%), papillary carcinomas (10%) and chromophobe carcinomas.

In addition, cytogenetic analysis has led to the discovery of new variants of renal cell carcinomas. In the 2004 WHO classification, a rare entity, "Xp11.2 translocation-related carcinoma", has appeared.

Several translocations involving the Xp11.2 region have been reported. These different translocations are grouped under the term MiTF/TFE translocation (1).

A few cases have been described in adults, but the frequency seemed to be underestimated in the absence of molecular biological research.

We report a rare case of carcinoma with Xp11.2 translocation revealed by hematuria and then discuss the epidemiological, diagnostic and therapeutic aspects through a review of the literature.

Case Présentation:

This is T.A aged 21, with no particular pathological history. The history of illness goes back 5 years with the appearance of a total intermittent clotting hematuria without any other associated sign. The whole evolving in a context of alteration of the general state. The clinical examination found a conscious patient, eupneic, with discolored conjunctiva. The urogenital examination found a lumbar tenderness with a positive left lumbar contact. The rest of the examination was unremarkable.

A biological check-up was requested showing an anemia of 6.5 g/dl, grouping: B+.

A normal renal function with a creatinemia of 11 mg /l

A radiological assessment (Uroscanner) was also requested, showing a large left renal tumor process occupying almost the entire kidney, estimated at 19.5*14*12. The tumor was found to be approximately 5 cm in diameter in the major axes, and appeared to be accompanied by an extension of the excretory cavities which appeared dilated and accompanied by nodular involvement of the peri-renal fat opposite the lower pole of the kidney in contact with the psoas muscle with multiple voluminous adenopathies predominantly retroperitoneal in the pre-aortic-cavity area, in the left latero-aortic area and at the level of the left renal hilum, these ADPs encompassing the renal vascular pedicle (Figure 1).

The patient underwent a packed red blood cell transfusion and then an enlarged total nephrectomy with a pre aortic-cavity lymph node curage. The postoperative course was simple.

After a multidisciplinary meeting, a PET scan was performed, which did not reveal any other secondary location. The patient presented a good clinical and biological evolution with no back pain or hematuria.

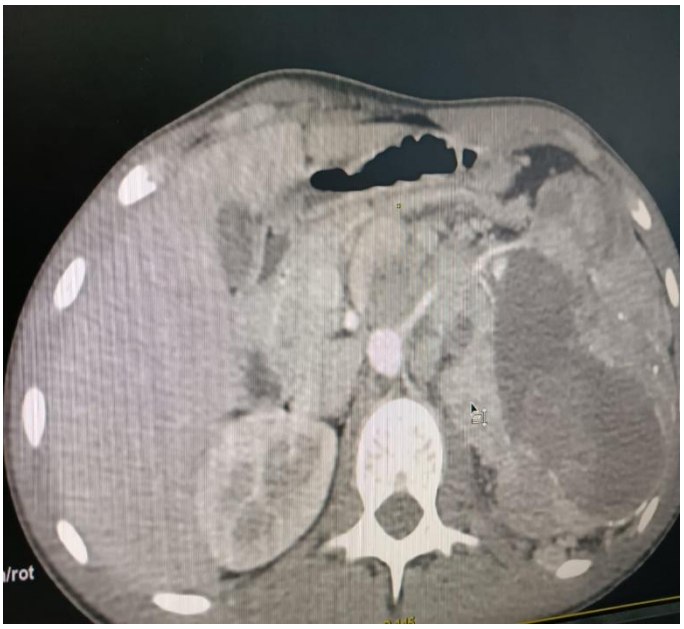

Figure 1 : Left renal tumor process estimated at 19.5*14*12.5 cm with extension to the excretory cavities and accompanied by nodular involvement in the peri-renal fat.

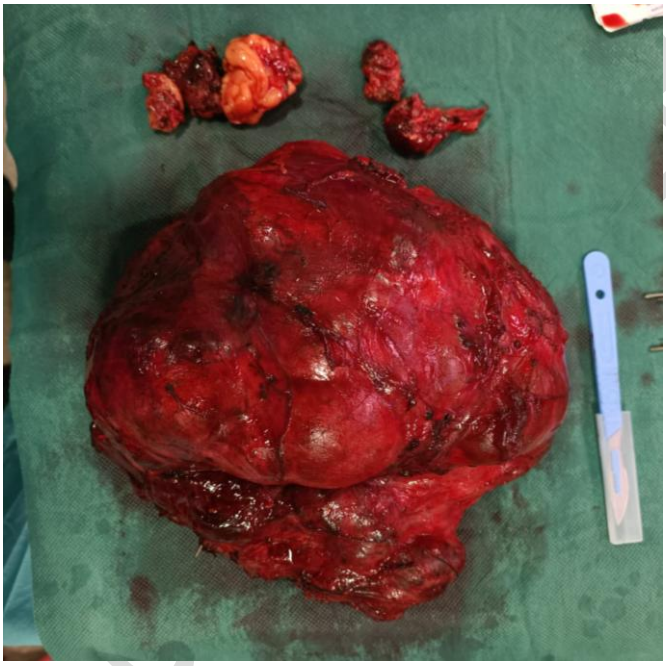

Figure 2 : An enlarged total nephrectomy with lymph node dissection extended to the aortic bifurcation

Discussion :

Initially observed preferentially in children and young adults and is characterized by a translocation involving band Xp11.2. This type, "Xp11 translocation CRC," has been recognized in the 2004 WHO classification. It can be confused with conventional cell carcinoma or tubulo-papillary carcinoma type 2. This translocation involves the TFE3 gene, at Xp11.2, encoding a transcription factor of the MiTF family.

There have been few cases described in the literature of translocation renal cell carcinoma in patients after 50 years of age, whereas series in children are numerous (2).

The incidence of translocation Xp11.2 renal cancer is low. Previous studies have reported an incidence of 0.9-5% of adult CRCs (3). Komai estimated its incidence in a series of 443 patients with renal cell carcinoma to be 1.6% and 15% in patients younger than 45 years (4).

In contrast to other types of kidney cancer, female gender is predominant in the literature (5).

Studies have found a history of chemotherapy in childhood in 15% of cases (6). In adults, the discovery is most often incidental. However, in young people under 30 years of age, there are often suggestive symptoms in two thirds of the cases diagnosed (7). Our 21-year-old patient presented with calliottent hematuria associated with chronic low back pain, which is consistent with the literature.

Microscopically they present as proliferations of variable architecture, papillary, alveolar or trabecular, made of tumor cells with cytoplasm

In the absence of a specific anatomopathological character, tRCCs pose a problem of differential diagnosis with other RCCs. The most common diagnostic method for Xp11.2 translocation RCC is the IHC test using an antibody to the C-terminal part of TFE3 (8)

In addition, genetic testing is the gold standard of diagnosis. It is requested in the first instance to allow a decision to be made after an equivocal immunohistochemical examination.

In our case, the histology was not sufficient to establish a diagnostic certainty: we observed renal cell carcinoma, then we completed by an immunohistochemical study which objectified a renal carcinoma with MITF family translocation

In the case of locally advanced tumors, the reference treatment is extended nephrectomy with removal of associated lymph nodes (9). In the literature, no case of neoadjuvant or isolated treatment by immunotherapy or anti angiogenic therapy has been described.

We performed an enlarged total nephrectomy with latero aortic lymph node curage. The postoperative course was simple.

The evolution of tRCC in adults is often negative, with a high risk of lymph node metastases. The response rate to targeted therapies is estimated at 30% with a survival of less than 2 years at the metastatic stage. While the prognosis remains favorable in children (10). The evolution of our patient was marked by a good clinical and biological improvement with absence of low back pain, hematuria and normalization of the hemoglobin level.

Conclusion :

Xp11.2 translocation carcinoma has been a very rare entity in adults

in adults. Its discovery appeared to be at a more advanced stage and its prognosis in adults was stage and its prognosis in adults was more negative than in other prognosis in adults was more negative than in other renal cell carcinomas.

The therapeutic management remained to be defined.

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