Original Article

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Clinical heterogeneity of Xp11 translocation renal cell carcinoma: impact of fusion subtype, age, and stage

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Xp11 translocation renal cell carcinomas harbor chromosome translocations involving the Xp11 breakpoint, resulting in gene fusions involving the TFE3 gene. The most common subtypes are the ASPSCR1-TFE3 renal cell carcinomas resulting from t(X;17)(p11;q25) translocation, and the PRCC-TFE3 renal cell carcinomas, resulting from t(X;1)(p11;q21) translocation. A formal clinical comparison of these two subtypes of Xp11 translocation renal cell carcinomas has not been performed. We report one new genetically confirmed Xp11 translocation renal cell carcinoma of each type. We also reviewed the literature for all published cases of ASPSCR1-TFE3 and PRCC-TFE3 renal cell carcinomas and contacted all corresponding authors to obtain or update the published follow-up information. Study of two new, unpublished cases, and review of the literature revealed that 8/8 patients who presented with distant metastasis had ASPSCR1-TFE3 renal cell carcinomas, and all but one of these patients either died of disease or had progressive disease. Regional lymph nodes were involved by metastasis in 24 of the 32 ASPSCR1-TFE3 cases in which nodes were resected, compared with 5 of 14 PRCC-TFE3 cases (P=0.02).; however, 11 of 13 evaluable patients with ASPSCR1-TFE3 renal cell carcinomas who presented with N1M0 disease remained disease free. Two PRCC-TFE3 renal cell carcinomas recurred late (at 20 and 30 years, respectively). In multivariate analysis, only older age or advanced stage at presentation (not fusion subtype) predicted death. In conclusion, ASPSCR1-TFE3 renal cell carcinomas are more likely to present at advanced stage (particularly node-positive disease) than are PRCC-TFE3 renal cell carcinomas. Although systemic metastases portend a grim prognosis, regional lymph node involvement does not, at least in short-term follow-up. The tendency for PRCC-TFE3 renal cell carcinomas to recur late warrants long-term follow-up.

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