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The concordant analysis of target gene sequencing data showing the tumor heterogeneity in triplet-paired metastatic tumor tissues in metastatic renal cell carcinoma

Eur Urol Suppl 2019; 18(1);e80

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Introduction & Objectives: The study aimed to analyze the inter/intratumoral concordance of expressed target genes from triple-paired metastatic tissues using target gene sequencing (TGS) in metastatic renal cell carcinoma (mRCC)

Materials & Methods: Between 2002 and 2017, a total of 350 triple-paired metastatic tissue samples from 262 mRCC patients obtained from either nephrectomy, or metastatectomy were used for target gene sequencing of 88 candidate genes from kidney cancer panel by the MACROGEN@, specialized for cancer gene sequencing. After quality check, a final 81 patients' 243 tissues were analyzed for TGS. The concordance of triple paired tissues were analyzed with 88 TGS by the bioinformatics.

Results: Among the 81 patients, only 49 (55.7%) patients had positive gene expression among 88 RCC cancer panel genes, whereas 38 (55.7%) patients had no pathogenic gene detection.As for the concordance of gene expressions between triple-paired tissues, only 12 patients had >95%of concordance among three tissues and 18 patients had >95% of concordance within two tissues and the rest of 8 patients' tissues had less than 50% of concordance. After all the consideration of genetic alterations including deletion, insertion, missense and nonsense mutation and splicing gene, the most frequently detected expression of genes were PTEN loss, followed by the FLCN, BCR, SMARCA2, AKAP9, MLH1, MYH11, APC and TP53.

Conclusions: The study showed the different genetic alterations affected by the organ sites and multi-heterogeneity of mRCC tissues. The concordance of pathogenic gene alteration detected within tissues was not high and even half of the patients had no pathogenic gene alterations.