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The Whole-Exon Gene Sequence Application in Clinical Rare Cancers

Ying Xu

Doctor of Surgery Postdoctoral Fellow in Oncology Specialty, China

In our clinical working, the multiple primary cancer (MPC) in a single patient occurred increasingly. These patients always possessed particular histopathology, physiology, and clinical symptoms, implicating that the normal treatments might not be suitable for them. To explore the tumorigenesis and prognosis of MPC, we utilized the novel whole-exon gene sequence assay to detect potential mutative genes based on paraffine tumor species from two patients: one of them was diagnosed as thyroid collision tumor (TCT), the other was diagnosed as renal cancer, breast cancer and thyroid papillary cancer. The result showed that a total of 76 single nucleotide variants (SNV), 12 insertion and deletion variants (INDEL), and 99 copy number variations (CNV) were identified in TCT, while only PCMTD with the copy number variation (CNV) was associated with the MPC patient. In summary, these clustered special genes in MPC and TCT disease give us new sight to deeply understand the tumorigenesis and prognosis of rare malignant tumours of patients.