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Genetic profiling of thymic carcinoma using targeted next-generation sequencing

Abstract

Thymic carcinoma is a rare mediastinal neoplasm and little is known about its tumorigenesis. There is no effective treatment except for complete resection, and the prognosis of advanced cases is poor. To identify the mutations associated with tumorigenesis, we analyzed genetic profile of thymic carcinoma using targeted next-generation sequencing. We sequenced about 409 cancer-related genes in 12 thymic squamous cell carcinoma tissues including 10 tumor / normal tissue pairs using Ion AmpliSeq Cancer Panel and Ion PGM Sequencer. We filtered the mutations with Ingenuity Variant Analysis, SIFT, PolyPhen-2, and PROVEAN. Twenty-five candidate mutations in 24 genes were identified, including five tyrosine kinase genes (*KIT, DDR2, PDGFRA, ROS1, IGF1R*). There was no recurrent mutation among the samples studied. The *KIT* exon 11 deletion mutation in 1 patient was an activating mutation and may be an oncogenic driver mutation. Genetic profiling of thymic carcinoma using targeted next-generation sequencing was performed. The mutation status of thymic squamous cell carcinoma is highly heterogeneous.