Whole Exome Sequencing of Primary Esophageal Squamous Cell Carcinoma and Esophageal Adenocarcinoma in One Individual

Xiaolong Tao, Xin Song, Ran Wang, Zongmin Fan, Xuena Han, Kan Zhong, Bei Li, Lidong Wang*

State Key Laboratory of Esophageal Cancer Prevention & Treatment and Henan Key Laboratory for Esophageal Cancer Research of the First Affiliated Hospital, Zhengzhou University, Zhengzhou, China

Email address:

xltao2000@126.com (Xiaolong Tao), xsong2012@126.com (Xin Song), wangran7758521@126.com (Ran Wang), zongminfan@126.com (Zongmin Fan), xuenahan@126.com (Xuena Han), zhongkan666@126.com (Kan Zhong), mbeili@126.com (Bei Li), ldwang2007@126.com (Lidong Wang)

*Corresponding author

Abstract

Objective To compare the genomic variants of primary esophageal squamous cell carcinoma (ESCC) and primary esophageal adenocarcinoma (EAC) using a special model of concurrent ESCC and EAC in the same individual. *Methods* Six samples of ESCC, EAC and normal esophageal tissues from two patients with concurrent ESCC and EAC in the same individual were subjected to whole-exome sequencing by using whole-exome sequencing. *Results* ESCC and EAC have different mutational features, and 19 mutated genes shared in esophageal squamous carcinoma and esophageal adenocarcinoma were found (OBSCN, TMEM261, ZNF462, STYXL1, EDEM3, SCN2A, WDR87, MACROD2, PAGE1, ANKRD18B, OR4Q3, GOLGA3, A2ML1, CACNA1A, PSMD3, CD1E, EPRS, PCDH11X and CHN1), three genes (DST, PCM1 and KIAA1328) were found to be mutated both in the two ESCC, and no co-occurring genes were found in the two EAC. *Conclusion* ESCC and EAC have obvious characteristics of tumor heterogeneity and gene mutation, and may have the same molecular mechanism.

Keywords

Concurrent Cancer, Simultaneity, Esophageal Squamous Cell Carcinoma, Esophageal Adenocarcinoma, Gene Mutation