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Genomic analysis reveals mutational signatures and frequently altered genes in esophageal squamous cell carcinoma

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Esophageal squamous cell carcinoma (ESCC) is the eighth most common and the sixth most lethal cancer worldwide. However, although genomic studies have identified some mutations associated with ESCC, we know little of the mutational processes responsible. To identify genome-wide mutational signatures, we performed either whole-genome sequencing (WGS) or whole-exome sequencing (WES) on 104 ESCC patients and combined our data with 88 samples reported previously. An APOBEC-mediated mutational signature in 47% of 192 tumors suggests that APOBEC-catalyzed deamination provides a source of DNA damage in ESCC. Moreover, PIK3CA hotspot mutations (c.1624G>A:p.Glu542Lys, c.1633G>A:p.Glu545Lys) were enriched in APOBEC-signature tumors and no smoking-associated signature was observed in ESCC. In the samples analyzed using WGS, we identified novel focal (<100 kb) amplifications of CBX4 and CBX8. In our combined cohort, we identified frequent inactivating mutations in AJUBA, ZNF750, PTCH1 and the chromatin-remodeling genes CREBBP and BAP1 in addition to known mutations. Functional analyses suggest roles for several genes (CBX4, CBX8, AJUBA and ZNF750) in ESCC. Notably, high activity of Hedgehog signaling and PI3K pathway in approximately 60% of 104 ESCC tumors indicates that therapies targeting these pathways may be particularly promising strategies for ESCC. Collectively, our data provide comprehensive insights into the mutational signatures of ESCC and identify new markers for early diagnosis and potential therapeutic targets.

Biography

Yongping Cui has completed her PhD from Cancer Institute and Cancer Hospital, Chinese Academy of Medical Sciences and Peking Union Medical College, China and Postdoctoral studies from H. Lee Moffitt Cancer Center, Tampa, FL, USA. She is the Director of Translational Medicine Research Center, Shanxi Medical University. She has published more than 21 papers in reputed journals and has been serving as an Editorial Board Member of repute.

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