Kai Ye, J Women's Health Care 2018, Volume 7
DOI: 10.4172/2167-0420-C2-010

Kai Ye
Xi'an Jiaotong University, China

Systematic discovery of complex insertions and deletions in human cancers

Complex insertions and deletions (indels) are formed by simultaneously deleting and inserting DNA fragments of different sizes at a common genomic location. Here we present a systematic analysis of somatic complex indels in the coding sequences of samples from over 8,000 cancer cases using Pindel-C. We discovered 285 complex indels in cancer-associated genes (such as PIK3R1, TP53, ARID1A, GATA3 and KMT2D) in approximately 3.5% of cases analyzed; nearly all instances of complex indels were overlooked (81.1%) or misannotated (17.6%) in previous reports of 2,199 samples. In-frame complex indels are enriched in PIK3R1 and EGFR, whereas frameshifts are prevalent in VHL, GATA3, TP53, ARID1A, PTEN and ATRX. Furthermore, complex indels display strong tissue specificity (such as VHL in kidney cancer samples and GATA3 in breast cancer samples). Finally, structural analyses support findings of previously missed, but potentially druggable, mutations in the EGFR, MET and KIT oncogenes. This study indicates the critical importance of improving complex indel discovery and interpretation in medical research.

Biography

Kai Ye has completed his PhD from Leiden University in the Netherlands and Postdoctoral studies from European Bioinformatics Institute in UK. He has worked as an Assistant Professor at Leiden University Medical Center and Washington University in St. Louis. He has published more than 50 papers in reputed journals.

kaiye@xjtu.edu.cn

Notes: