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생명정보학, 유전체학, 체성돌연변이 분석

[논문]

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Exploring the role of somatic mutations in human brain diseases

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With advances in next-generation sequencing (NGS) technologies, somatic mutation analysis has made great strides in establishing a comprehensive description of genomic changes in cancer over the past decade. International collaborative efforts such as The Cancer Genome Atlas (TCGA) and International Cancer Genome Consortium (ICGC) have been made to construct the landscape of somatic mutations in various cancer types, leading to unprecedented knowledge of the somatic mutations and their underlying mechanisms.

Recent analyses of somatic mutations have begun to be made in non-cancerous diseases, especially in brain diseases. Unlike cells in other tissues, differentiated neurons in the brain are rarely replaced and regenerated during the course of a person's life, so that somatic mutations in those cells may critically affect the function of neurons and even brain

circuits. Recent studies have demonstrated that somatic mutations occurring during brain development do actually cause multiple neurodevelopmental diseases, supporting a new pathogenesis of brain disorders. However, due to the lack of specialized bioinformatic tools for detecting rare somatic mutations in tissues without clonal expansion, somatic mutations in brain have not been comprehensively explored yet.

In this talk, the speaker will discuss the difficulties associated with analyzing somatic mutations in the brain, as well as the current efforts aimed at tackling these challenges. The talk will cover different sequencing approaches and developed bioinformatic methods used to detect rare somatic mutations in low cell population or even in a single cell.