

## 2023년 제 3회 통계세미나

고려대학교 통계연구소와 BK21 통계학교육연구팀, 그리고 DS+가 다음과 같이 공동으로 세미나를 개최하오니 많은 참여 바랍니다.

일시 : 2023년 3월 24일(금) 오전 11시

장소 : 고려대학교 정경관 206 호

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### Scalable statistical test for identifying disease-associated variants in regulatory DNA

<Abstract>

Mutations in the noncoding DNA, which represents approximately 99% of the human genome, have been crucial to understanding disease mechanisms through dysregulation of disease-associated genes. One key element in gene regulation that noncoding mutations mediate is the binding of proteins to DNA sequences. Genetic variants such as single nucleotide polymorphisms, and insertion and deletion of bases (InDels) may impact on the gene regulation and downstream biological processes through disruption or creation of protein-DNA binding. We develop novel tests of statistical significance using a Markov model to evaluate the impact on the binding affinity and identify genetic variants altering protein-DNA binding. The test predicts binding changer variants of regulatory significance with an efficient importance sampling algorithm generating background sequences in favor of large binding affinity changes. The application to human leukemia data uncovers candidate pathological variants on modulating binding of important transcription factors in leukemic patients.

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