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# Fundamental and applied 3D genomics

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This year marks the 15th year of the pioneering publication that introduced a genome-wide chromosome conformation capture technique, setting a milestone in 3D genomics. Over the past decade and a half, these techniques have become essential tools for addressing a wide array of genomic challenges. Our session on "Fundamental and Applied 3D Genomics" will explore key questions in this dynamic field. Initially, we will explore recent discoveries concerning the mechanisms of chromatin organization. Studies of our group, utilizing genetically modified mouse models, have elucidated a complex relationship between gene expression regulation and the maintenance of local chromatin domains, emphasizing the role of CTCF in this intricate process. Contrarily, we will also present cases from specific cell types demonstrating that CTCF binding does not always correlate with chromatin domain formation. Additionally, we introduce new physical models that illuminate how active transcription influences the formation of these CTCF-independent structures. Our findings significantly enhance our understanding of 3D genomics and its implications for regulatory mechanisms underlying human monogenic diseases and cancer development.

Another key focus of our research involves the application of chromatin conformation technologies to detect structural variants and improve genome assembly. We will highlight innovative computational methods and experimental workflows that facilitate the detection of both structural and single-nucleotide variants in the human genome. These methods have proven crucial in cancer research and the molecular diagnosis of monogenic diseases. Furthermore, the advent of single-cell techniques has enabled the application of these technologies to low-input human samples, ultimately making them suitable for analyzing human embryo biopsies. These advancements allow for the effective identification of balanced chromosomal translocations in clinical settings.

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