5 DECEMBER

13.30 P.M.
ROOM B108
POVO 2

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BEYOND THE CODING DNA: UNCOVERING REGULATORY MUTATIONS

Despite the integration of next-generation sequencing technologies in clinical practice, approximately 50% of patients with rare genetic disorders (RGDs) remain without a definitive diagnosis, resulting in significant challenges for patients and their families. This diagnostic gap likely arises from multiple factors, but a substantial portion of the "missing heritability" is thought to reside in non-coding and regulatory variants, which are often overlooked or not thoroughly evaluated in routine clinical settings.

This talk aims to present illustrative cases of RGDs associated with regulatory variants that disrupt proper gene transcription. Specifically, we will examine how Hi-C technology can enhance the diagnostic rate for RGDs by enabling the characterization of regulatory variants that alter the 3D conformation of the genome and the structure of topologically associating domains (TADs). The identification and clinical interpretation of regulatory variants located within the non-coding genome (98% of DNA) represent a central challenge for human genetics in the coming decades. Addressing this challenge could significantly improve the diagnostic rate for currently undiagnosed patients with rare genetic disorders, ultimately ending the "diagnostic odyssey" for patients and their families.

CIBIO EXTERNAL SEMINAR

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