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# Computational prediction of disease effects of genetic variants and altered 3D chromosome folding patterns

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**Abstract:** With the rapid progress of genome studies, many missense single-nucleotide polymorphisms (SNPs) in populations of somatic cells of different diseases such as cancer have been identified. In addition, 3D chromosome studies are emerging as a major source of information on transcription machineries important for disease development. However, it is challenging to identify and understand the implications of disease-related variants. In addition, deciphering the relationship between disease associated variants and 3D chromosome folding is still in its infancy. Here we describe recent progress in developing computational methods to assess the effects of missense mutations and chromosome folding of genomic regions enriched with cancer-variants. By integrating protein sequence information, structural and topological properties of protein conformations, we show machine-learning based predictions of pathological effects of mutations can be made, with improved performance compared to current state-of-the-art methods on challenging data sets. We discuss how key relevant residues, including previously unreported cancer-variants, can be identified from a large number of background variants found in cancer patients. Furthermore, we discuss how cancer-related variants can be uncovered through identification of higher-order cooperative units of clusters of residues that function collectively and cooperatively. Finally, we discuss insight gained from deciphering the folding mechanism of 3D genome, and results on differential patterns of chromatin folding of genomic regions enriched in cancer variant mutations.

**Biosketch:** Dr. Jie Liang is Richard and Loan Hill professor in the Dept of Bioengineering at the University of Illinois at Chicago (UIC), and UIC Distinguished Professor. He received BS (Biophysics, 1986) from Fudan University, MCS and Ph.D. (Biophysics, 1994) from the University of Illinois at Urbana-Champaign. He was an NSF CISE postdoctoral fellow (1994-1996) at the Beckman Institute and National Center for Supercomputing and its Applications (NCSA). He was a fellow at the Institute of Mathematics and Applications (IMA) at Minneapolis during 1996-97, an Investigator at SmithKline Beecham Pharmaceuticals during 1997-98. He joined UIC in 1999 and became a full professor in 2007. He was a recipient of the NSF CAREER award (2003), a fellow of the American Institute of Medicine and Biological Engineering (2007), and a University Scholar (2010). Dr. Liang's research interests include structural bioinformatics, 3D chromosome, molecular stochastic networks, cellular pattern formation, and topological data analysis. His recent work can be found at ([gila.bioe.uic.edu/liang/liang\\_pub.html](http://gila.bioe.uic.edu/liang/liang_pub.html)).

Talk recording:

<https://pitt.hosted.panopto.com/Panopto/Pages/Viewer.aspx?id=17b0a737-3899-4221-bfd4-add0011f3430>