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3D genome organization and the genetic risk to (some) diseases

Genome-wide association studies have established statistical associations between various diseases and a large number of single-nucleotide polymorphisms (SNPs). However, they provided no simple explanation of the mechanisms underlying this association. Following the idea that 3D genome organization and its variations may have a functional role in gene regulation, we investigated the distribution of these SNPs with respect to specific 3D features, first focusing on topologically-associating domains (TADs) and their borders. Our computational analyses have shown that for some specific diseases, including many cancers, disease-associated SNPs are over-represented in TAD borders. To analyze further this enrichment, its determinants and its consequences, we have selected candidate loci and started an experimental study using two techniques (HRS-seq and 3C-qPCR) developed in the group at IGMM.

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