سمینار هفتگي ماده چگال نرم

# عنوان سمینار

## Inter-chromosomal hubs and disruption of chromatin organization in mouse model of Autism Spectrum Disorder (ASD)-linked 16p11.2 CNV


ارائه دهنده

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**چکیده**

Structural variants in the human 16p11.2 locus spanning a 600kb region with 29 genes are associated with early-onset autism spectrum disorder with variable degree of penetration. To investigate the changes in transcriptional regulation that accompany the complex phenotypes observed in the presence of 16p11.2 structural variants, we have measured gene expression and chromatin folding in mouse embryonic stem cells and during in vitro differentiation into neuronal progenitor cells and early dopaminergic neurons. We are also analyzing matched Hi-C data to ask whether the changes in expression are due to altered chromatin contacts. Using polymer physics and computer simulations, we connect our findings to topological features of 16p11.2 locus and its inter-chromosomal contacts. Our findings may provide novel insights into the critical, yet understudied mechanisms underlying the damaging role of the 16p11.2 variant for the neuronal development.


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