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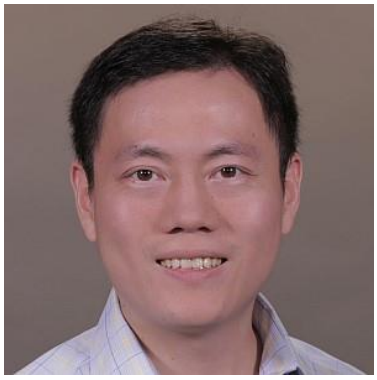
The Human Genetics Institute of New Jersey
Collaborative Research for Translational Solutions

DEPARTMENT OF GENETICS
&
HUMAN GENETICS INSTITUTE OF NEW JERSEY

RESEARCH SEMINAR

"Characterization of repeat expansion disorders from long-read sequencing"



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Short tandem repeats (STRs) are directly adjacent repetitions of specific nucleotide motifs in a genome. Here we first describe computational approaches to quantify repeats within STRs. In addition, long-read sequencing can also be used to characterize the haplotype structure of the region encompassing STRs. We demonstrate its use on a 10kb region in exon-1 of HTT, which causes Huntington's disease (HD). One SNP common to 30% of individuals with HD of European ancestry was identified for allele-specific CRISPR/Cas9 deletion of the expanded repeats in human HD cell lines. Our workflow can be applied to other repeat expansion diseases.

Noon, Monday, December 5, 2022
Auditorium, Life Sciences Building,
145 Bevier Road, Busch Campus, Piscataway, New Jersey
Zoom Registration Link | Meeting ID: 963 3843 9086 |Passcode: Genetics

Host: Jinchuan Xing | Phone: 848-445-9663 | Email: jinchuan.xing@rutgers.edu