

# Child Health and Human Development Program



## MONDAY NOON SEMINARS

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**October 18, 2021, 12-1 PM**

## Splicing alterations associated with disease variants

### Seminar Highlights

- Disease-associated variants can occur in splicing regulators and cis-acting elements that result in aberrant RNA splicing
- Long-read sequencing technology allows us to identify the full complexity of aberrant RNA
- A remaining challenge in the field is understanding the functional consequences of aberrant RNA splicing

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