

DBMI OPEN INSIGHTS

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DEPARTMENT OF BIOMEDICAL INFORMATICS
HARVARD MEDICAL SCHOOL

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Dissecting Germline and Somatic Mutational Processes Across Space and Time



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I will present a neural network approach to mutational signature decomposition. Using this method, we elucidated the origins of clock-like signatures and uncovered strong associations between tumor hypoxia and defective DNA repair. We further identified a germline mutational hotspot at transcription start sites enriched in mosaic variants, suggesting an origin in early embryogenesis. (Full abstract and bio online.)

Friday, March 7, 2025 • 3:00–4:00pm • Countway Library • Lahey Room (5th Floor)

The **DBMI Open Insights Seminars** are occasional research talks related to the mission of the Department of Biomedical Informatics. This includes themes such as:

- Provisioning big data to the scientific community
- Getting the big picture on human health
- Learning from each patient
- Advancing basic science with data science
- Understanding disease beyond heredity: environmental impact
- Instrumenting the health enterprise for discovery and intervention



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