

Reference Materials and Benchmarks for Reproducible Genomics: Lessons from ‘Genome In A Bottle’



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2:00 -3:00pm ET

<https://rocku.zoom.us/j/93659505719?pwd=SGFkKa5TczEJRhiRkRWZR1g1NAV2Qk.1>



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ABSTRACT: How do we know whether a genomic result is correct and reproducible? As genome sequencing technologies and analysis methods rapidly evolve, well-characterized reference materials and benchmark datasets provide a common foundation for understanding their strengths, limitations, and sources of disagreement. The NIST Genome in a Bottle Consortium brings together government, academic, and industry researchers to develop openly available reference materials, benchmark datasets, and benchmarking methods for human genome sequencing.

The talk will discuss lessons from Genome in a Bottle about designing and using benchmarks to improve rigor and reproducibility in genomics. It will highlight how complementary sequencing technologies and analysis methods can be integrated to establish high-confidence benchmarks, how benchmarking can reveal systematic errors and difficult regions of the genome, and why clearly defining what is and is not covered by a benchmark is essential for interpreting performance results. The talk will also discuss recent efforts to extend benchmarking toward increasingly complete genomes and more challenging applications, including cancer genomics, and remaining opportunities for improving reproducibility as genomic technologies continue to advance.

Speaker’s Bio: Dr Justin Zook, co-leads the Biomarker and Genomic Sciences Group at the National Institute of Standards and Technology (NIST) and the Genome in a Bottle Consortium, an open collaboration among government, academia, and industry developing benchmark reference materials and data for human genome sequencing. His research focuses on integrating and comparing sequencing technologies to characterize genetic variation and developing benchmarks that enable researchers to evaluate the accuracy and reproducibility of genome sequencing and analysis methods. He received the Presidential Early Career Award for Scientists and Engineers (PECASE) for his work in this area.