CANSSI ONTARIO **STAGE**

Heidi Rehm

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Global Strategies to Decipher Rare Disease

Friday
Jan 9, 2026
12–1 pm ET

Hybrid Event I Register Here
Rooms 10031 & 10032
10th Floor, 700 University Avenue
Toronto, ON

Abstract: Despite major technological advances in genomic sequencing and data analysis, the majority of individuals with a suspected genetic disorder do not receive a molecular diagnosis. So where is the gap and how do we address it? This talk will present strategies to identify novel causes of rare disease including the application of new technologies and analysis methods as well as building innovative approaches to global data sharing.

Profile: Heidi Rehm is Director of the Genomic Medicine Unit in the Center for Genomic Medicine and Chief Genomics Officer at Massachusetts General Hospital, working to integrate genomics into medical practice. She is an ABMGG-certified Clinical Laboratory Director of Broad Clinical Labs working to guide clinical genomic testing in medical practice. She is a principal investigator of ClinGen, providing free and publicly accessible resources to support the interpretation of genes and variants. Rehm also co-leads the Broad Center for Mendelian Genomics focused on discovering novel rare disease genes. She is a strong advocate and pioneer of open science and data sharing, working to extend these approaches through her role as Chair of the Global Alliance for Genomics and Health. Rehm is also a principal investigator of the Genome Aggregation Database (gnomAD) and the Broad All of Us Genome Center, generating genomic resources to fuel discovery. She serves as an advisor to the Clinical Pharmacogenetics Implementation Consortium, Danish National Genome Center, Monarch Initiative, CIViC database and Ensembl. She received the Scientific Achievement Award from the American Society of Human Genetics in 2022 and the Coulter Award from the Association for Diagnostics and Laboratory Medicine in 2025.

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