

CAGI7 Conference

The Seventh Edition of the Critical Assessment of Genome Interpretation

Final Program



Saturday
6 December 2025

Northeastern University, EXP Building, 8th Floor
815 Columbus Avenue, Boston, MA 02115

8:30-9:00 **Registration & Breakfast**

9:00-9:30 **Welcome, Perspective, and Charge to Participants**

9:30-11:00 **ARSA Missense Challenge**

- 9:30-9:50 Introduction by Steven Brenner, University of California, Berkeley
- 9:50-10:10 Dean Suhr, patient advocate, MLD Foundation
- 10:10-10:30 (D) Michael Gelb, University of Washington
- 10:30-10:50 (A) Jennifer Lin, University of California, Berkeley
- 10:50-11:00 Discussion

11:00-11:15 **Break**

11:15-12:00 **Keynote** Heidi Rehm, Massachusetts General Hospital and Broad Institute

12:00-1:00 **Lunch**

1:00-1:25 **Rare Genomes Project**, Anne O'Donnell-Luria, Broad Institute of MIT and Harvard

1:25-2:25 **TSC2 Missense Challenge**

- 1:25-1:30 Introduction
- 1:30-1:45 (D) Doug Fowler, University of Washington
- 1:45-2:05 (A) Vikas Pejaver, Mount Sinai School of Medicine
- 2:05-2:15 (P) Kyungkeon Chung, UX Factory
- 2:15-2:25 Discussion

2:25-3:10 **Distinctive Advances in Missense Prediction**

- 2:25-2:45 (P) Fabrizio Pucci, Université Libre de Bruxelles
- 2:45-3:00 (P) Gianluca Lombardi, Sorbonne Université
- 3:00-3:10 (P) *Mustafa Tekpinar, Van Yüzüncü Yıl University

3:10 – 3:25 **Break**

3:25–5:00 **FGFR Missense Gain-of-Function and Pharmacogenetic Challenge**

- 3:25-3:30 Introduction
- 3:30-3:50 (D) Sven Diederichs, Albert-Ludwigs-University Freiburg & DKTK
- 3:50-4:15 (A) *Russ Altman, and Helix Group Assessors, Stanford
- 4:15-4:30 (P) Doyeon Ha, 3billion
- 4:30-4:45 (P) Kerr Ding, Georgia Institute of Technology

- 4:45-5:00 Discussion

5:00-5:30 **Annotation Accumulation Accuracy Assessment (Annotate All) Challenges**

- 5:00-5:10 Missense variants: Predrag Radivojac, Northeastern University
- 5:10-5:20 In-frame indels: (A) Haneen Abderrazzaq, Northeastern University
- 5:20-5:30 Nonsense & frameshifting variants: (A) Yaqi Su, University of California, Berkeley

5:30-6: **Selected Talks**

- 5:30-5:40 (P) Alexander Gress, Helmholtz Institute
- 5:40-5:50 (P) Hoyin Chu, Memorial Sloan Kettering Cancer Center
- 5:50-6:00 (P) Yihong Yang, Texas A&M University

Dinner on Own

Sunday
7 December 2025

Northeastern University, East Village, 17th Floor
291 St. Botolph Street, Boston, MA 02115

8:30-9:00 **Registration & Breakfast**

- Posters up, please arrive at 8:15am

9:00-10:40 **ATP7B & BARD1 Missense Challenges**

- 9:00-9:05 Introduction
- 9:05-9:20 (D) Fritz Roth, University of Pittsburgh Medical Center
- 9:20-9:35 (D) Lea Starita, University of Washington
- 9:35-10:05 (A) *Emidio Capriotti, University of Bologna
- 10:05-10:15 (P) Panagiotis Katsonis, Baylor College of Medicine
- 10:15-10:25 (P) Brynja Matthiasardottir, University of Maryland and NIH
- 10:25-10:40 Discussion

10:40-10:55 **Break**

10:55-11:40 **Keynote** Doug Fowler, University of Washington

11:40-12:00 **Ethics Forum** Flavia Chen, UT Southwestern

12:00-1:00 **Lunch**

1:00-1:30 **Missense Challenges Discussion**

- 1:00-1:30 Sean Mooney, CIT, NIH, discussion leader

1:30-2:50 **lentiMPRA Expression Regulation Challenge**

- 1:30-1:35 Introduction
- 1:35-1:50 (D) Ryan Hernandez, UCSF
- 1:50-2:10 (A) Michael Beer, Johns Hopkins University
- 2:10-2:20 (P) *Dmitry Penzar, Lomonosov Moscow State University
- 2:20-2:30 (P) Arif Rather, Boston Children's Hospital
- 2:30-2:40 (P) John Butts, The Jackson Laboratory
- 2:40-2:50 Discussion

2:50-3:05 **Break**

3:05-4:00 Polygenic Risk Scores Challenge

- 3:05-3:10 Introduction
- 3:10-3:25 (D) Shamil Sunyaev, Harvard Medical School
- 3:25-3:45 (A) Sung Chun, Harvard Medical School
- 3:45-3:55 (P) Doug Speed, Aarhus University
- 3:55-4:00 Discussion

4:00-4:25 Advancing Genome Interpretation Ryan Hernandez, UCSF

4:25-5:05 The Future of CAGI

- 4:25-4:45 Doug Speed, Aarhus University
- 4:45-5:05 Discussion

5:05-5:45 Selected Talks

- 5:05-5:15 Laura Covill, Broad Institute of MIT and Harvard
- 5:15-5:25 Timothy Bergquist, Icahn School of Medicine at Mount Sinai
- 5:25-5:35 Lindsay Romo, Broad Institute of MIT and Harvard
- 5:35-5:45 Peiran Jiang, Carnegie Mellon University

5:45-7:30 Reception & Posters

Monday
8 December 2025

Northeastern University, East Village, 17th Floor
291 St. Botolph Street, Boston, MA 02115

8:30-9:00 Registration & Breakfast

- Posters down

9:00-10:10 Splicing Challenge

- 9:00-9:15 Introduction by Steve Mount, University of Maryland
- 9:15-9:30 (D) Kinga Bujakowska, Harvard Medical School
- 9:30-9:55 (A) Sirui Zhang, Harvard Medical School
- 9:55-10:10 Discussion

10:10-11:10 LPL Missense Challenge

- 10:10-10:15 Introduction
- 10:15-10:30 (D) Daniel Tabet, Broad Institute of MIT and Harvard
- 10:30-10:50 (A) Joe Marsh, University of Edinburgh
- 10:50-11:05 (P) *Shaopei Ye, Vall d'Hebron Institut de Recerca

11:05-11:15 Concluding Remarks

11:15-12:30 Assessor Office Hours

(A) Assessor
(D) Data provider
(P) Predictor
