CAGI✸ Workshop 2019
Assessing the Future of Genome Interpretation

6 – 8 December 2019

Agenda

Friday, 6 December 2019

12:00-1:00 pm **Registration & light refreshments**

1:15-3:10 pm **Session 1 – Predicting risk: from Mendelian to polygenic traits**

1:15-2:00 pm **Shamil Sunyaev**
Predicting polygenic risk

2:00-2:30 pm **Melissa Cline**
CAGI Theme: CAGI cancer challenges

2:30-3:10 pm **Selected talks / flash talks**

Lipika Ray (15 minutes)
Evolution of CAGI over four rounds: computational challenges in analyzing next generation sequencing data

Natàlia Padilla Sirera (5 minutes)
BRCA1- and BRCA2-specific in silico tools for variant interpretation in the CAGI 5 ENIGMA challenge

Andrew Sharo (5 minutes)
StrVCTVRE: A supervised learning method to predict the pathogenicity of structural variants

Kymberleigh Pagel (5 minutes)
OpenCRAVAT: an open source collaborative platform for the annotation of human genetic variation

Vikas Pejaver (5 minutes)
A performance-based approach to establish standards for missense

Yue Cao (1 minute)
Predicting pathogenicity of missense variants with weakly supervised regression

Justin Delano (1 minute)
The impact of missense human variation on post-translational modifications in proteins
variant impact prediction tools

3:10-3:40 pm ***Break***

3:40-5:00 pm **Session 2 – Ethical considerations for CAGI**

3:40-3:55 pm **Selected talk**
Zhiqiang Hu
Privacy time bombs in omics data: latent risk manifests over time

3:55-4:35 pm **Barbara Koenig and Malia Fullerton**
The CAGI Ethics Forum

4:35-5:00 pm Discussion

5:00-5:30 pm ***Break***

5:30-6:30 pm **Session 3 – Reports from the CAGI Salon: inspirations for the future**

5:30-5:40 pm **Steven Brenner**

Overview, and charge to participants

5:35-5:45 pm **Vikas Pejaver**
CAGI Salon introduction

5:45-6:00 pm **Kymberleigh Pagel**
CAGI challenge design and assessment

6:00-6:15 pm **Lipika Ray**
Broadening impact and participation in CAGI

6:15-6:30 pm Discussion

6:30-9:00 pm ***Reception*** **and Poster session**

Saturday, 7 December 2019

8:00-8:45am ***Breakfast***

8:45-9:00 am **Welcome and Recap from Day 1**

9:00-10:45 am **Session 4 – Broader perspectives on Human Genome Variation**

9:00-9:45 am **Michael Snyder**
Big data and health

9:45-10:45 am **Resources and policies**

9:45-10:15 am **Anne O'Donnell-Luria**
gnomAD

10:15-10:45 am **Marc Greenblatt**
ClinGen Sequence Variant Interpretation

10:45-11:15 am ***Break***

11:15-1:00 pm **Session 5 – Missense variants: promises and limitations**

11:15-11:45 am **John Moult**
CAGI Theme: Bespoke approaches often enhance performance with biophysical methods excelling in a few cases while evolutionary methods have a more consistent performance

11:45-12:15 pm **Iddo Friedberg**
CAGI Theme: Prediction methods have high statistical significance but accuracy is low. However, for an identifiable subset of predictions, accuracy is very high

12:15-12:45 pm **Olivier Lichtarge**
CAGI Theme: Methods tend to correlate with each other more than with experiment

12:45-1:00 pm CAGI Theme Discussion

1:00-2:00 pm ***Lunch***

2:00-4:00 pm **Session 6 – Complex traits and non-coding variants**

2:00-2:30 pm **Sean Mooney**
CAGI Theme: Predicting complex traits from exomes is fraught, although there have been improvements in the ability to match genomes to profile

2:30-3:00 pm **Steve Mount**
CAGI Theme: There have been improvements in splicing prediction although this is not yet at the state of missense

3:00-3:15 pm Discussion

3:15-3:45 pm **Predrag Radivojac**
Overcoming (many) challenges in evaluating variant interpretation: a machine learning perspective

3:45-4:00 pm Discussion

4:00-4:40 pm ***Break***

4:30-5:30 pm **Session 7 – Assessment of the annotate all missense CAGI challenge**

4:30-5:00 pm **Nilah Ioannidis,** assessor

5:00-5:30 pm Discussion

5:30-6:30 pm **Meta-predictor panel – potential, prevarication, and policy**
Panelists: Rachel Karchin, Nilah Ioannidis, Sean Mooney
Moderator: Steven Brenner

Sunday, 8 December 2019

8:00-8:45am ***Breakfast***

8:45-9:00 am **Welcome and Recap of Day 2**

9:00-10:30 am **Session 8 – AI and diagnostic variants**

9:00-9:45 am **Serafim Batzoglou**
AI methods for genome interpretation

9:45-10:15 am **Constantina Bakolitsa**
CAGI Theme: CAGI challenges have led to the identification of causal variants overlooked by clinical labs

10:15-10:30 am **Selected talk**
Erwin Frise
Benchmarking an artificial intelligence method for fast diagnosis of rare genetic disease

10:30-11:00 am ***Break***

11:00-12:00 pm **Session 9 – Future perspectives**

11:00-11:45 am **Future of genome interpretation and vision for CAGI panel**
Panelists: Rachel Karchin, Olivier Lichtarge, Vikas Pejaver
Moderator: Steven Brenner

11:45-12:00 pm **Final remarks**