



Translational Bioinformatics Conference 2017



Friday – September 29, 2017

7:00 am - 8:00 am – Breakfast and Registration

8:00 am - 8:15 am – Welcome and Introductions – Dr. Dokyoon Kim & Dr. Marylyn Ritchie

8:15 am - 9:00 am – Keynote: Dr. David Ledbetter, *Precision Health at Geisinger: Longitudinal electronic health data and exome sequence on 250K participants*

9:00 am - 9:45 am – Keynote: Dr. Jessie Tenenbaum, *Translational Bioinformatics: Past, Present, and Future*

9:45am - 10:15 am – Break

Precision medicine pipelines and applications

Session Chair: Dr. Dokyoon Kim

10:15 am-10:35 am – (P) Local genetic ancestry in CDKN2B-AS1 is associated with primary open-angle glaucoma in an African American cohort extracted from de-identified electronic health records (Nicole Restrepo)

10:35 am- 10:50 am – (A) Identification of disease associations with rare Loss of Function variants from 50,000 exomes in Geisinger Health System (Anurag Verma)

10:50 am - 11:05 am - (A) iDEG: A single-subject method utilizing two-paired transcriptomes of an individual to compute differentially expressed genes (Qike Li)

11:05am - 11:25 am - (P) Rare variants in the Splicing Regulatory Elements of EXOC3L4 are associated with brain glucose metabolism in Alzheimer's disease (Jason Miller)

11:25 am - 11:40 am – (A) A genome-by-environment interaction classifier for precision medicine: personal transcriptome response to rhinovirus identifies children prone to asthma exacerbations (Yves Lussier)

11:40 am - 12:00 pm - (P) Frequency and phenotype consequence of APOC3 rare variants in patients with very low triglyceride levels (Dana Crawford)

12:00 pm-1:00 pm – **Lunch**

Pharma Informatics using the EHR

Session Chair: Dr. Kye Hwa Lee

1:00 pm-1:45 pm – Keynote: Dr. Nicholas Tatonetti, *Observational data for biomedical discovery*

1:45 pm-2:00 pm – (A) Depression diagnosis and antidepressant treatment associated with contemporary prescriptions of narcotics and benzodiazepines with worse outcomes (Wendy Marie Ingram)

2:00 pm-2:20 pm – (P) Pharmacological Risk Factors Associated with Hospital Readmission Rates in a Psychiatric Cohort Identified using Prescriptome Data Mining (Shameer Khader)

2:20 pm-2:40 pm – (P) City-wide Analysis of Drug-Drug-Interactions (Luis Rocha)

2:40 pm-3:00 pm – **Break**

3:00 pm-3:45 pm – Keynote: Dr. Ju Han Kim, *Precision dinner: a personal interpretation of pharmaco and metabolic genomes*

Flash poster talks

Session Chair: Dr. Dokyoon Kim

3:45 pm-3:53 pm – Using a simulation approach to evaluate data-driven algorithms for studying clinical heterogeneity in complex traits (Anna Basile)

3:53 pm-4:01 pm – Probabilistic graphical model of ICD-9 codes from electronic health records (EHRs) of Geisinger Health System (GHS) patients for disease risk prediction conditioned on prior medical history (Marta Byrska-Bishop)

4:01 pm-4:09 pm – Anti-diabetic Rosiglitazone Regulates mRNA Stability of Lipolytic Genes (Kyoung Jae Won)

4:09 pm-4:17 pm – CAS-viewer: Web-based analysis and visualization of alternative splicing in cancer (Younghee Lee)

4:17 pm-4:25 pm – Heterogeneous Network Based Rare Disease Gene Prioritization (Aditya Rao)

4:25 pm-4:33 pm – Challenges and approaches of systems neurobiology networks to understand mechanisms and genetic risk of Alzheimer's disease (Yuan R. Shang)

4:33 pm-4:41 pm – Collective feature selection to identify important variables for epistatic interactions: A simulation study (Shefali Verma)

4:41 pm-4:49 pm – Computational analyses of single-subject 'omics to develop a 'personalome': How far are we from clinically-interpretable results? (Francesca Vitali)

5:00 pm-6:30 pm – Poster Session

6:30 pm – Dismissal – **Dinner** on your own

Saturday – September 30, 2017

7:00 am - 8:00 am – **Breakfast**

Medical informatics and text mining in electronic health records

Session Chair: Dr. Indira Ghosh

8:00 am-8:45 am – Keynote: Dr. Dana Crawford, *Diversity in precision medicine research: an example in Cleveland*

8:45 am-9:00 am – (A) Extracting metadata from large biomedical data repositories and mapping to the bioCADDIE metadata specification DATS (Data Tagging Suite) (Nansu Zong)

9:00 am-9:15 am – (A) Latent-based imputation of laboratory measures from Electronic Health Records (V. Abedi)

9:15 am-9:35 am – (P) Comparison of MetaMap and cTAKES for entity extraction in clinical notes: Some remarks about aggregation and semantic types (Ruth Reategui)

9:35 am-9:55 am – (P) Text Mining MEDLINE for Rare Disease Gene Prioritization (Aditya Rao)

9:55 am-10:15 am – **Break**

Sequencing analysis pipelines for efficient computing

Session Chair: Dr. Yonghee Lee

10:15 am-10:30 am–(A) More Complete Variant Discovery in Large Exome Sequencing Projects by Optimization of Analytical Pipelines (Shulan Tian)

10:30 am-10:45 am–(A) Parallel distributed PCR duplication marking algorithm integrated with genome sequence alignment by using streaming technology (Junehawk Lee)

Across the transcriptome – methods and applications

Session Chair: Dr. Yonghee Lee

10:45 am-11:00 am– (A) Power Analysis of summary based methods for identifying expression-trait associations (Yogasudha Veturi)

11:00 am-11:20 am – (P) A novel joint analysis framework improves identification of differentially expressed genes in cross disease transcriptomic analysis (Wenyi Qin)

11:20 am-11:40 am – (P) How does normalization matter in RNA-seq disease diagnosis? (Henry Han)

11:40 am-12:00 pm– (P) Indel sensitive and comprehensive variant/mutation detection from RNA sequencing data for precision medicine (Sun Zhifu)

12:00 pm-1:00 pm – **Lunch**

Machine learning technologies for bioinformatics

Session Chair: Dr. Luis Rocha

1:00 pm-1:45 pm – Keynote: Dr. Jason Moore, *Artificial intelligence for everyone*

1:45 pm-2:05 pm – (P) Deep learning predicts breast cancer estrogen receptor status from metabolomics data (Fadhl Alakwaa)

2:05 pm-2:25 pm – (P) Clinical Opinion Generation from General Blood Test Results Using Binary Relevance Neural Network with Opinion Grouping (Youjin Kim)

2:25 pm-2:45 pm – (P) Min-Redundancy and Max-Relevance Multi-view Feature Selection for Predicting Ovarian Cancer Survival using Multi-omics Data (Yasser El-Manzalawy)

2:45 pm-3:00 pm – **Break**

Applications of network and pathway analysis to understand disease

Session Chair: Dr. Vida Abedi

3:00 pm-3:45 pm – Keynote: Dr. Ida Sim, *Personal Mobile Technologies for Precision Medicine*

3:45 pm-4:05 pm– (P) Pathway Networks Generated from Human Disease Phenome (Ann Cirincione)

4:05 pm-4:25 pm– (P) Integrative Pathway based Survival Prediction utilizing Interaction between Gene Expression and DNA Methylation in Breast Cancer (So Yeon Kim)

4:25 pm-4:40 pm–(A) Survival analysis identifies germline variants associated with overall survival among an endometrial cancer cohort (Jason Miller)

4:40 pm-5:25 pm – Keynote: Dr. Atul Butte, *Translating Trillions of Points of Data into Therapies, Diagnostics, and New Insights into Disease*

5:25 pm-5:45 pm – Closing Remarks/Best Paper & Abstract Award

5:45 pm – Dismissal

6:00 pm – Banquet

Sunday – October 1, 2017

8:00 am – 9:00am - Tutorial: Dr. Marylyn Ritchie, *Precision Medicine: Study Design, Tools, and Applications*

9:00am - 10:00am – Tutorial: Dr. Dokyoon Kim, *Multi-omics Data Integration for Translational Bioinformatics*

10:00 am – Closing remarks and TBC 2017 Concludes!

(P): Paper talk, (A): Abstract talk