#### **PACIFIC SYMPOSIUM ON BIOCOMPUTING 2017**

2017 marks the 22nd Pacific Symposium on Biocomputing (PSB)! Biocomputing, biomedical informatics and data science have become high profile activities in recent years. The associated emphasis on the need for policies and technologies for effective data sharing has also received quite a bit of attention. In a series of editorials, the New England Journal of Medicine (Longo & Drazen, N Engl J Med 2016; 374:276-277. 1/21/2016. DOI: 10.1056/NEJMe1516564 and Drazen, N Engl J Med 2016; 374:e24. 5/12/2016. DOI: 10.1056/NEJMe1601087) discussed the particular challenges for data sharing in the context of clinical data, generating much discussion about the proper approaches for these activities. Most notable, however, was the introduction of the phrase "research parasite" to describe "people who had nothing to do with the design and execution of the study but use another group's data for their own ends, possibly stealing from the research productivity planned by the data gatherers, or even use the data to try to disprove what the original investigators had posited." Of course, this perfectly describes (although in somewhat more negative terms than are typically used by data scientists) the motivation behind data sharing in genomics and molecular biology-and the potential value of secondary analysis of public data sets, including the occasional disproof of an incorrect scientific finding or conclusion—thus saving time, resources and potentially lives. The depiction of secondary analyses and the potential refutation of primary analyses as a negative surprised many, but points to the special challenges of sharing sensitive clinical data. Nonetheless, "I am a research parasite" became a refrain among biomedical data analysts who felt strongly that these analyses are indeed quite positive and exactly what is needed. In that vein, the PSB organizers were approached with an idea to host an award to recognize those who have made substantial scientific contributions by analyzing data collected by others. These "Research Parasite Awards" quickly attracted financial support and will be presented for the first time at this meeting. To his credit, Dr. Jeffrey Drazen, the Editor-in-Chief of the New England Journal of Medicine, has also agreed to give a talk about the challenges in clinical data sharing, particularly from his vantage point as an editor of an influential clinical journal. Dr. Drazen has no connection with the Research Parasite Awards, but is fully aware that they have been created and will be awarded at the meeting this year.

The mission of PSB is to provide a forum for the best *emerging* science in Biocomputing, providing both formal and informal mechanisms for scientific communication. PSB depends on the community to define emerging areas in biomedical computation. Its sessions are usually conceived at the previous PSB meeting as people discuss trends and opportunities for new science. The typical program includes sessions that evolve over two to three years as well as entirely new sessions. This year we revisit new dimensions of precision medicine, ranging from single cell measurements to populations.

In addition to being published by World Scientific and indexed in PubMED, the proceedings from all PSB meetings are available online at <u>http://psb.stanford.edu/psb-online/</u>. PSB has published more than 800 papers. These papers are often cited in journal articles and represent early contributions in emerging subfields—many times before there is an established literature in more traditional journals; for this reason, many papers have garnered hundreds of citations. The Twitter handle PSB 2017 is @PacSymBiocomp and the hashtag this year will be #psb17.

The efforts of a dedicated group of session organizers have produced an outstanding program. The sessions of PSB 2017 and their hard- working organizers are as follows:

### Computational approaches to understanding the evolution of molecular function

Yana Bromberg, Matthew Hahn, and Predrag Radivojac

#### **Imaging Genomics**

Li Shen and Lee Cooper

#### Methods to Ensure the Reproducibility of Biomedical Research

Konrad J. Karczewski, Nicholas P. Tatonetti, Chirag J. Patel, Arjun K. Manrai, C. Titus Brown, and John P.A. Ioannidis

# Patterns in Biomedical Data - How do we find them?

Anurag Verma, Anna Okula Basile, Marta Byrska-Bishop, Christian Darabos, H. Lester Kirchner, and Sarah Pendergrass

# Precision medicine: from genotypes and molecular phenotypes towards improved health and therapies

Bruce Aronow, Steven E. Brenner, Dana C. Crawford, Joshua C. Denny, Sean D. Mooney, and Alexander A. Morgan

### Single-cell analysis and modelling of cell population heterogeneity

Nikolay Samusik, Sean Bendall, and Nima Aghaeepour

We are also pleased to present four workshops in which investigators with a common interest come together to exchange results and new ideas in a format that is more informal than the peer-reviewed sessions. For this year, the workshops and their organizers are:

# Harnessing Big Data for Precision Medicine: Infrastructures and Applications

Kun-Hsing Yu, Steven Hart, Rachel Goldfeder, Qiangfeng Cliff Zhang, Stephen Parker, and Michael Snyder

# The Making of Next Generation Data Scientists in Biomedicine

Lana Garmire, Shamim Nemati, John D. Van Horn, Jason Moore, Carole Shreffler and Michelle Dunn

**No-Boundary Thinking in Bioinformatics** 

Xiuzhen Huang and Jason H. Moore

# **Open Data for Discovery Science**

Philip R.O. Payne, Kun Huang, Nigam H. Shah, and Jessica Tenenbaum

We thank our keynote speakers Neil Risch (Science keynote) and David Magnus (Ethical, Legal and Social Implications keynote). We also thank Jeffrey Drazen for his talk.

Tiffany Murray has managed the peer review process and assembly of the proceedings since 2003, and also plays a key role in many other aspects of the meeting. We are grateful for the support of the The Penn Institute for Biomedical Informatics; Rxight Pharmacogenetics Program; and the Institute for Computational Biology, a collaborative effort of Case Western Reserve University, the Cleveland Clinic Foundation, and University Hospitals for their support of PSB 2017. We also thank the National Institutes of Health<sup>1</sup> and the International Society for Computational Biology (ISCB) for travel grant support. The research parasite awards benefit by support from: Jeff Stibel, GigaScience (Biomed Central), Nature Genetics, Scientific Data (Nature), the Gordon & Betty Moore Foundation, the Arnold Foundation, Tim Triche Jr., and Casey Greene.

We are particularly grateful to the onsite PSB staff Al Conde, Ryan Whaley, Georgia Hansen, BJ Morrison-McKay, Cynthia Paulazzo, Jackson Miller, Kasey Miller, and Paul Murray for their assistance. We also acknowledge the many busy researchers who reviewed the submitted manuscripts on a very tight schedule. The partial list following this preface does not include many who wished to remain anonymous, and of course we apologize to any who may have been left out by mistake.

<sup>&</sup>lt;sup>1</sup> Funding for this conference was made possible (in part) by Grant # 5 R13 LM006766 – 20 from the National Library of Medicine. The views expressed in written conference materials or publications, and by speakers and moderators, does not necessarily reflect the official policies of the Department of Health and Human Services; nor does mention by trade names, commercial practices, or organizations imply endorsement by the U.S. Government.

We look forward to a great meeting once again. Aloha!

Pacific Symposium on Biocomputing Co-Chairs, October 15, 2016

**Russ B. Altman** Departments of Bioengineering, Genetics & Medicine, Stanford University

**A. Keith Dunker** Department of Biochemistry and Molecular Biology, Indiana University School of Medicine

Lawrence Hunter Department of Pharmacology, University of Colorado Health Sciences Center

Marylyn D. Ritchie Department of Biomedical and Translational Informatics, Geisinger Health System

**Teri E. Klein** Department of Genetics, Stanford University

#### Thanks to the reviewers...

Finally, we wish to thank the scores of reviewers. PSB aims for every paper in this volume to be reviewed by three independent referees. Since there is a large volume of submitted papers, paper reviews require a great deal of work from many people. We are grateful to all of you listed below and to anyone whose name we may have accidentally omitted or who wished to remain anonymous.

Nima Aghaeepour Harindra Arachchi Mohammad Arbabshirani Bruce Aronow Chloe-Agathe Azencott Anna Basile Kayhan Batmanghelich Chris Bauer Asa Ben-Hur Sean Bendall Tyler Burns William Bush Mariusz Butkiewicz Fabien Campagne Kevin Chen Shuo Chen Yin Hoon Chew Moo Chung Jessica Cooke Bailey James Costello Christian Darabos Christophe Dessimoz Lei Du Todd Edwards Arne Elofsson Niclas Eriksson Tilman Flock Yi Gao Brice Gaudilliere Tian Ge Jeff Gentry Olivier Gevaert Jesse Gillis Anthony Gitter Rachel Goldfeder Casey Greene Jake Hall Xiaoke Hao Yun Hao Imran Haque Jaroslaw Harezlak Blanca Himes Isaac Hodes **Emily Holzinger** Ting Hu Junzhou Huang Jake Hughev Shaun Jackman Ola Jacunski

Shuiwang Ji Kipp Johnson Konrad Karczewski Jonathan Karr Dokvoon Kim Sungeun Kim H. Lester Kirchner Jun Kong Linglong Kong Mickey Kosloff Irina Kufareva Willaim Lai Chirag Lakhani Nicholas Larson Roman Laskowski LiLi Ruowang Li Nita Limdi Jingyu Liu Kefei Liu Liang Liu Colton Llovd Tal Lorberbaum Jose Lugo-Martinez Gang Luo Emily Mallory Elisabetta Manduchi Ariun Manrai Tyler Massaro Brett McKinnev Andrew Michaels Marghoob Mohiyuddin Jason Moore Yves Moreau Spencer Muse Kelly Nudelman Randy Olson Casey Overby Bernhard Palsson Gaurav Pandev Chirag Patel Vikas Pejaver Sarah Pendergrass Hanchuan Peng Minoli Perera Abhishek Pratap Wei-Oi Oei Marylyn Ritchie Igor Rogozin

Joe Romano Mert Sabuncu Satya Sahoo Erin Simonds Marina Sirota Johannes Soding Sudeep Srivastava Jason Stein Timothy Sweeney Suzanne Tamang Haixu Tang Nicholas Tatonetti Shaolei Teng Gregg Thomas William Thompson Stefano Toppo Ryan Urbanowicz Giorgio Valentini Elise Valkanas Sofie Van Gassen Fabio Vandin Shankar Vembu Anurag Verma Yogasudha Veturi Bjarni Vihjalmsson Susann Vorberg Pei Wang Qianghu Wang Marquitta White Chunlei Wu Rong Xu Ya Yang Dmeliha Yetisgen Pooya Zakeri Daogiang Zhang Xiaobo Zhou Chengsheng Zhu