Long-Read
Sequencing Workshop

The Jackson Laboratory
for Genomic Medicine
Farmington, Conn.

May 16 - 20, 2022

JAX **The Jackson Laboratory**

*Leading the search
for tomorrow's cures*

www.jax.org/longread

Welcome

Dear Friends and Colleagues,

It is our pleasure to welcome you to the third annual Long Read Sequencing Workshop hosted by The Jackson Laboratory for Genomic Medicine in Farmington, Connecticut. This year, we aim to bring together experts in genetics, technology development, data science and medicine to share the latest advances in the field of long-read sequencing.
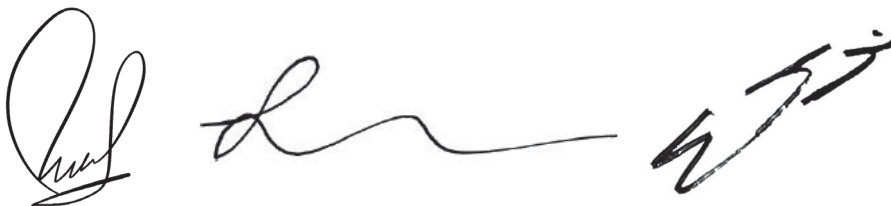
Our vision for this event is to serve as a forum for technology developers and users, academic researchers and industry representatives to exchange new ideas and push the boundaries of the state-of-the-art long-read sequencing technologies. We further hope that this workshop serves as an educational platform to foster the development of the next generation of genome scientists – proving access to the most up-to-date technologies, protocols and analytic tools.

This year, our scientific program is anchored by three days of featured lectures highlighting exciting developments and novel discoveries achievable by leveraging long-read sequencing technologies. On the fourth day, the focus of the lectures will be on computational approaches used to effectively analyze long-read sequencing data and the state-of-the-art tools being implemented by bioinformaticians today. Finally, our program will conclude with an optional day of hands-on wet-lab tutorials, showcasing current laboratory techniques to generate long-read sequencing libraries and data.

We want to take this opportunity to thank the many people who made this workshop a reality – our esteemed speakers for their valuable time and expertise, our sponsors for their generous support, The Jackson Laboratory's Education and Genome Technologies teams, and last but not the least, your participation which makes this workshop possible.

We look forward to five days of exceptional tutorials and exciting discussions!

With best wishes,

Pamela Goldaraz, Tina Beaulieu and Chia-Lin Wei
Coordinating Committee

Long-Read
Sequencing Workshop

May 2022

Long-read sequencing technologies have emerged as powerful players in genomics over the last few years.

These new technologies have proven their ability to resolve some of the most challenging regions of genomes, leading to, among other things, the discovery of novel mechanisms of disease.

The workshop's five-day program includes three days of plenary sessions featuring renowned speakers, expert users, and technology developers who will share their experiences applying long-read sequencing in basic and translational genomic science and also explore the commonalities and differences in the current technologies. The workshop also offers one day of hands-on wet-lab tutorial and an additional day of sessions covering computational approaches for effectively analyzing long-read sequencing data with state-of-the-art tools.

Throughout the event, participants will have the opportunity to network with genomics experts and industry leaders.
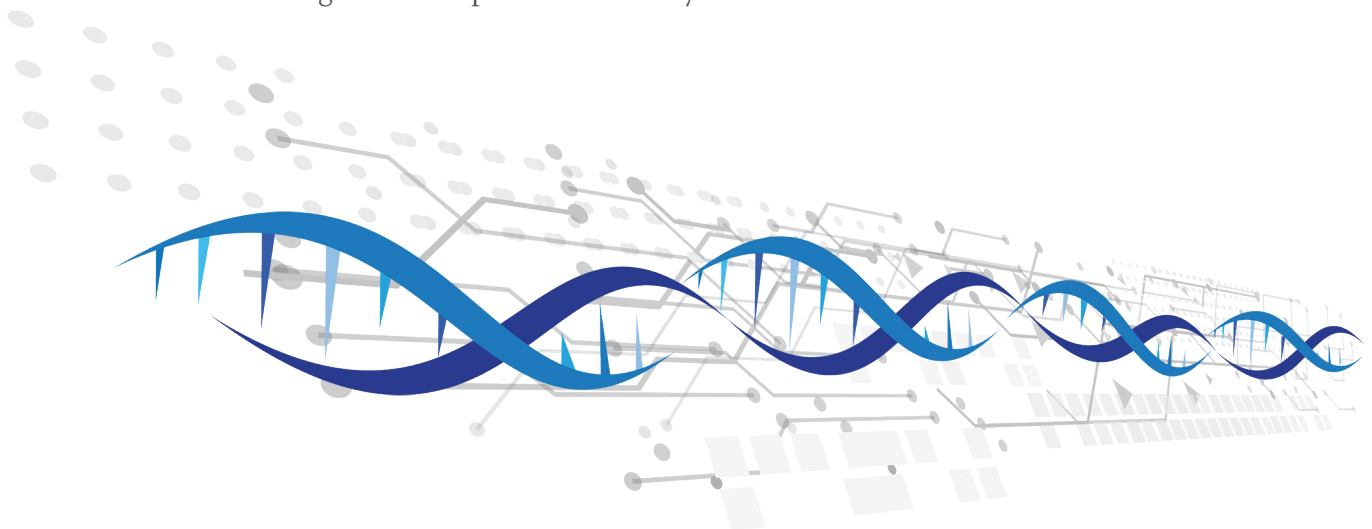
Agenda

Day 1 – Monday, May 16, 2022

8:00 a.m. Registration & COVID-19 on-arrival testing

Welcome
9:15 a.m. **Chia-Lin Wei, Ph.D.**, Professor & Director of Genome Technologies, The Jackson Laboratory

9:20 a.m. **The Past, Present, and Future of Sequencing Technologies in Genomic Medicine**
    George Weinstock, Ph.D., Professor, Evnin Family Chair and Director of Microbial Genomics,
        The Jackson Laboratory

10:00 a.m. Coffee Break

Session 1:   Technologies
10:30 a.m. **Realizing High Throughput, Human Whole Genome Sequencing**
    Kelvin Liu, Ph.D., Vice President of Technology Development, Pacific Biosciences

11:00 a.m. **DNA analysis using high-speed atomic force microscopy**
    Jason Reed, Ph.D., Professor, Virginia Commonwealth University

11:30 a.m. **Generating long read information without long-read sequencing**
    Tom Chen, Ph.D., Co-Founder & CSO, Universal Sequencing Technology

12:00 p.m. Lunch

Session 2: Structural Variation and Cancer
1:30 p.m. **EccDNAs are apoptotic products with high innate immunostimulatory activity**
    Yi Zhang, Ph.D., Professor, Harvard Medical School

2:00 p.m. **Ultra-low error synthetic long-read sequencing to characterize human cancer**
    Jianhua Luo, Ph.D., Professor of Pathology, University of Pittsburgh

2:30 p.m. **Discovery of cancer neoantigens using long-read transcriptome sequencing**
    Wigard Kloosterman, Ph.D., Chief Scientific Officer, Frame Therapeutics & Cyclomics

3:00 p.m. Coffee Break

Session 3: Structural Variation and Cancer II

3:30 p.m. **EagleC: A deep-learning framework for predicting structural variations from bulk and single-cell Hi-C maps**
Feng Yue, PhD., Duane and Susan Burnham Professor of Molecular Medicine, Director of the Center for Cancer Genomics of Robert H. Lurie Comprehensive Cancer Center, Northwestern University

4:00 p.m. **Introducing CGLR: Cancer Genomics with Long Reads.**
Michael Schatz, Ph.D., Adjunct Professor of Quantitative Biology, Cold Spring Harbor Laboratory

4:30 p.m **Deciphering extrachromosomal circular DNA by long-read sequencing**
Chia-Lin Wei, Ph.D., Professor & Director of Genome Technologies, The Jackson Laboratory

Keynote

5:00 p.m. **Discovering new biology and function in complete, telomere-to-telomere (T2T) genomes**
Karen Miga, Ph.D., Assistant Professor and Associate Director, UCSC Genomics Institute

6:00 p.m. Welcome Reception / Cocktail Hour

Day 2 – Tuesday, May 17, 2022

Keynote

9:00 a.m. **Long-read sequencing, structural variation and disease**
Evan Eichler, Ph.D., Principal Investigator & Professor of Genome Sciences, University of Washington

10:00 a.m. Coffee Break

Session 4: Human Genome

10:30 a.m. **Comprehensive Structural Variant Detection: From Mosaic to Population-Level**
Fritz Sedlazeck, Ph.D., Associate Professor, Baylor College of Medicine

11:00 a.m. **Complete, telomere-to-telomere assembly of diploid genomes**
Adam Phillippy, Ph.D., Senior Investigator, National Human Genome Research Institute

11:30 a.m. **T2T-level genomics reveals the activity and diversity of human repetitive sequences**
Rachel O'Neill, Ph.D., Professor and Director, Molecular and Cell Biology, Institute Systems Genomics, University of Connecticut

12:00 p.m. Lunch – Optional Sponsor talk (Leo Holt Conference Room)

Biology at true resolution: Resolving complex biology at the single-cell level
Patrick Murphy, Ph.D., 10X Genomics

Agenda

Day 2 – Tuesday, May 17, 2022

Session 5: Epigenomics

1:30 p.m. **Mapping different forms of DNA methylation from bacteria, microbiome and human**
Gang Fang, Ph.D., Associate Professor, Icahn School of Medicine at Mount Sinai

2:00 p.m. **Using nanopore sequencing to interrogate the genome and epigenome**
Winston Timp, Ph.D., Associate Professor of Biomedical Engineering, Johns Hopkins University

2:30 p.m. **Single-molecule chromatin fiber sequencing**
Andrew Stergachis, MD, Ph.D., Assistant Professor of Medicine and Genome Sciences, University of Washington

3:00 p.m. Coffee Break

Session 6: Transcriptomics I

3:30 p.m. **RNA splicing alterations in breast aging and cancer**
Olga Anczuków, Ph.D., Associate Professor, The Jackson Laboratory

4:00 p.m. **Single-cell long-read transcript sequencing with scCOLOR-seq**
Adam Cribbs, Ph.D., Group Leader in Systems Biology and Next Generation Sequencing Analysis, The University of Oxford

4:30 p.m. **Investigate the complexity of transcriptome and epigenetics by single-molecule long-read sequencing**
Kin Fai Au, Ph.D., Associate Professor, Department of Biomedical Informatics, The Ohio State University

5:30 p.m. Poster Session & Sponsor Exhibition – Wine and cheese

Day 3 – Wednesday, May 18, 2022

Session 7: Plant Characterization

9:00 a.m. **Portable genomics, Social Justice and Saving Cassava**
Laura Boykin Okalebo, Ph.D., Senior Scientific Consultant, The BioTeam, Inc.

10:00 a.m. **Lowering STEM barriers with HiFi assembly of college campus tree genomes**
Alex Harkess, Ph.D., Faculty Investigator, Auburn University, Hudson Alpha Institute for Biotechnology

10:30 a.m. **Genome assembly and comparative evolutionary analysis of maize and its wild relatives**
Matthew Hufford, Ph.D., Associate and Cassling Family Professor, Iowa State University

11:00 a.m. Coffee Break

Session 8: Transcriptomics II

11:30 a.m. Nanopore sequencing of full-length circular RNA in brain reveals unique splicing patterns
Jørgen Kjems, Ph.D., Professor, Aarhus University

12:00 p.m. Mammalian brain isoforms at single-cell resolution
Hagen Tilgner, Ph.D., Assistant Professor, Weill Cornell Graduate School

12:30 p.m. Lunch – Optional Training Session (Leo Holt Conference Room)

Grant writing: Specific Aims Page Workshop
Carmen Robinett, Ph.D., Senior Scientific Writer, The Jackson Laboratory

Session 9 – Selected Abstracts talks

2:30 p.m. Generating Population-scale Long-read Sequencing from 4000 human brain samples to catalog structural variants in Alzheimer's disease and related Dementias: a CARD-NIH initiative
Kimberley J Billingsley, Ph.D., Center for Alzheimer's and Related Dementias, National Institute on Aging

2:50 p.m. Detection and assembly of extrachromosomal DNA in tumors with nanopore sequencing
Sergey Aganezov, Ph.D., Oxford Nanopore Technologies

3:10 p.m. Long range sequence analysis of the mutational burden of Transposable Elements and Endogenous Retroviruses in laboratory mice.
Peter Warburton, Ph.D., Icahn School of Medicine at Mount Sinai

3:30 p.m. Coffee Break

Session 10: Vertebrate Genome

4:00 p.m. Towards high-quality reference assemblies for all vertebrate genomes
Olivier Fedrigo, Ph.D., Director, Vertebrate Genome Laboratory, The Rockefeller University

4:30 p.m. Long Read Sequencing Reveals Extensive Structural Variation in Diverse Mouse Genomes
Christine Beck, Ph.D., Assistant Professor, The Jackson Laboratory

Closing Keynote

5:00 p.m. Long-reads for saving endangered species and discovering new ones
Christopher Mason, Ph.D., Professor, Weill Cornell Medicine

5:40 p.m. Closing Remark - Dinner

Agenda

Day 4 – Thursday, May 19, 2022
Computational Biology - Bioinformatics showcase sessions

Session 11: Basic long-read sequencing QC (Nanopore and PacBio)
9:00 a.m. **Where do I start? Fundamental analyses of Nanopore and PacBio sequencing data**
> **Gabriel Rech, Ph.D.,** Bioinformatics Analyst II, The Jackson Laboratory

Session 12: Metagenomic assemblies
9:45 a.m. **Long-read metagenomic assembly with metaFlye**
> **Mikhail Kolmogorov, Ph.D.,** Stadtman Investigator, Cancer Data Science Laboratory, NIH, Center for Cancer Research, National Cancer Institute

10:30 a.m Coffee Break (COVID testing)

Session 13: Bionano data analysis
11:30 a.m. **Bionano Access: Genome Visualization and Analysis**
> **Benjamin Clifford, Ph.D.,** Senior Field Application Scientist, Bionano Genomics

Session 14: Genome assembly
12:15 p.m. **Shasta: a de novo assembler for long reads**
> **Sara Simmonds, Ph.D.,** Product Applications Scientist, Chan Zuckerberg Initiative

1:00 p.m. Lunch

Session 15: Variants detection
2:00 p.m. **PRINCESS: A Framework for Comprehensive Detection of Haplotype Resolved SNVs, SVs, and Methylation**
> **Medhat Mahmoud, Ph.D.,** Postdoctoral Associate, Baylor College of Medicine

Session 16: Transcriptomics
2:45 p.m. **Tools for transforming long reads into functional insights about isoform expression**
> **Ana Conesa, Ph.D.,** Research Professor, Spanish National Research Council

3:30 p.m. Coffee Break

Session 17: Methylation detection
4:00 p.m. **Detecting methylation using nanopore direct DNA sequencing**
> **Sheng Li, Ph.D.**, Associate Professor, The Jackson Laboratory

Session 18: Taxonomic profiling
4:45 p.m. **Best practices for taxonomic profiling of long read shotgun metagenomics datasets**
> **Daniel Portik, Ph.D.,** Senior Bioinformatics Scientist, PacBio

5:30 p.m. Closing Remark

Day 5 – Friday, May 20, 2022
Hands-on wet-lab tutorial (Optional)

8:00 a.m Registration opens

8:30 a.m. Introduction
Rahul Maurya, M.S., The Jackson Laboratory

9:00 a.m. Extracting High Molecular Weight DNA from Cells
Meihong Li, Ph.D., The Jackson Laboratory

11:15 a.m. Lunch – Sponsor talk (Leo Holt Conference Room)
Femto Pulse instrument and HMW DNA QC
Kyle Luttgeharm, Ph.D., Agilent Technologies, Inc

12:30 p.m. PacBio HiFi SMRTBell Library Prep using Express Template Kit
Padmaja Ghospurkar, Ph.D., The Jackson Laboratory

2:30 p.m. Oxford Nanopore Targeted Library Prep and Sequencing (Part 1)
Christine Goldfarb, Ph.D., The Jackson Laboratory

3:00 p.m. Coffee Break

3:15 p.m. Oxford Nanopore Targeted Library Prep and Sequencing (Part 2)
Christine Goldfarb, Ph.D., The Jackson Laboratory

5:30 p.m. Wrap Up and Q&A

Speakers

Adam Cribbs, Ph.D.
Group Leader in Systems Biology and Next
Generation Sequencing Analysis
The University of Oxford

Adam Phillippy, Ph.D.,
Senior Investigator. National Human Genome
Research Institute

Alex Harkess, Ph.D.
Faculty Investigator. Auburn University,
Hudson Alpha Institute for Biotechnology

Ana Conesa, Ph.D.
Research Professor,
Spanish National Research Council

Andrew Stergachis, M.D., Ph.D.
Assistant Professor of Medicine and Genome
Sciences, University of Washington

Benjamin Clifford, Ph.D.
Senior Scientist, Clinical and Scientific Affairs,
Bionano Genomics, Inc.

Carmen Robinett, Ph.D.
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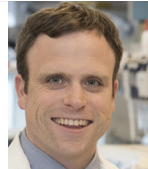
Christopher Mason, Ph.D.
Professor, Weill Cornell Medicine

Daniel Portik, Ph.D.
Senior Scientist, Bioinformatics, PacBio

Evan Eichler, Ph.D.
Principal Investigator & Professor of Genome
Sciences, University of Washington

Feng Yue, Ph.D.
Duane and Susan Burnham Professor of Molecular
Medicine, Director of the Center for
Cancer Genomics of Robert H. Lurie
Comprehensive Cancer Center,
Northwestern University

Fritz Sedlazeck, Ph.D.
Associate Professor, Baylor College of Medicine

Gabriel Rech, Ph.D.
Bioinformatics Analyst II, The Jackson Laborator

Gang Fang, Ph.D.
Associate Professor, Icahn School of Medicine
at Mount Sinai

George Weinstock, Ph.D.
Professor, Evnin Family Chair and Director of
Microbial Genomics, The Jackson Laboratory

Hagen Tilgner, Ph.D.
Assistant Professor, Weill Cornell
Graduate School

Jason Reed, Ph.D.
Professor, Virginia Commonwealth University

Jianhua Luo, Ph.D.
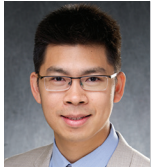Professor of Pathology, University of Pittsburgh

Jørgen Kjems, Ph.D.
Professor, Aarhus University

Karen Miga, Ph.D.
Assistant Professor and Associate Director,
UCSC Genomics Institute

Kelvin Liu, Ph.D.
Vice President of Technology Development,
Pacific Biosciences

Kin Fai Au, Ph.D.
Associate Professor, Department of Biomedical
Informatics, The Ohio State University

Laura Boykin Okalebo, Ph.D.
Senior Scientific Consultant, The BioTeam, Inc.

Matthew Hufford, Ph.D.
Associate and Cassling Family Professor,
Iowa State University

Medhat Mahmoud, Ph.D.
Postdoctoral Associate, Baylor College of Medicine

Michael Schatz, Ph.D.
Adjunct Professor of Quantitative Biology,
Cold Spring Harbor Laboratory

Mikhail Kolmogorov, Ph.D.
Stadtman Investigator, Cancer Data Science
Laboratory. NIH, Center for Cancer Research,
National Cancer Institute

Olga Anczuków, Ph.D.
Associate Professor, The Jackson Laboratory

Olivier Fedrigo, Ph.D.
Director, Vertebrate Genome Laboratory,
The Rockefeller University

Rachel O'Neill, Ph.D.
Professor and Director, Molecular and Cell Biology,
Institute Systems Genomics.
University of Connecticut

Sara Simmonds, Ph.D.
Product Applications Scientist,
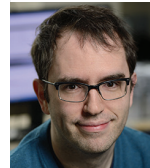Chan Zuckerberg Initiative

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Universal Sequencing Technology

Wigard Kloosterman, Ph.D.
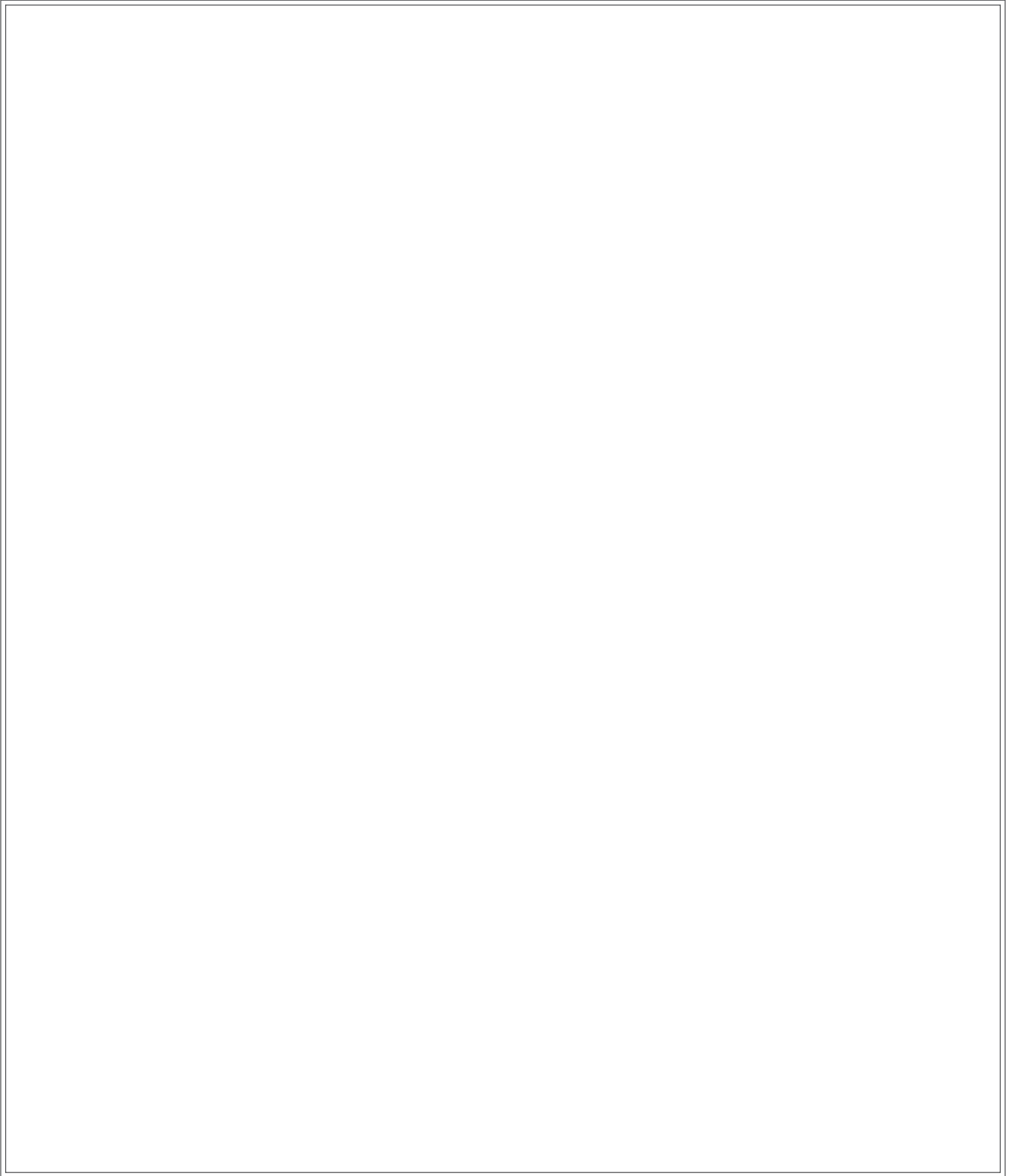Chief Scientific Officer,
Frame Therapeutics & Cyclomics

Winston Timp, Ph.D.
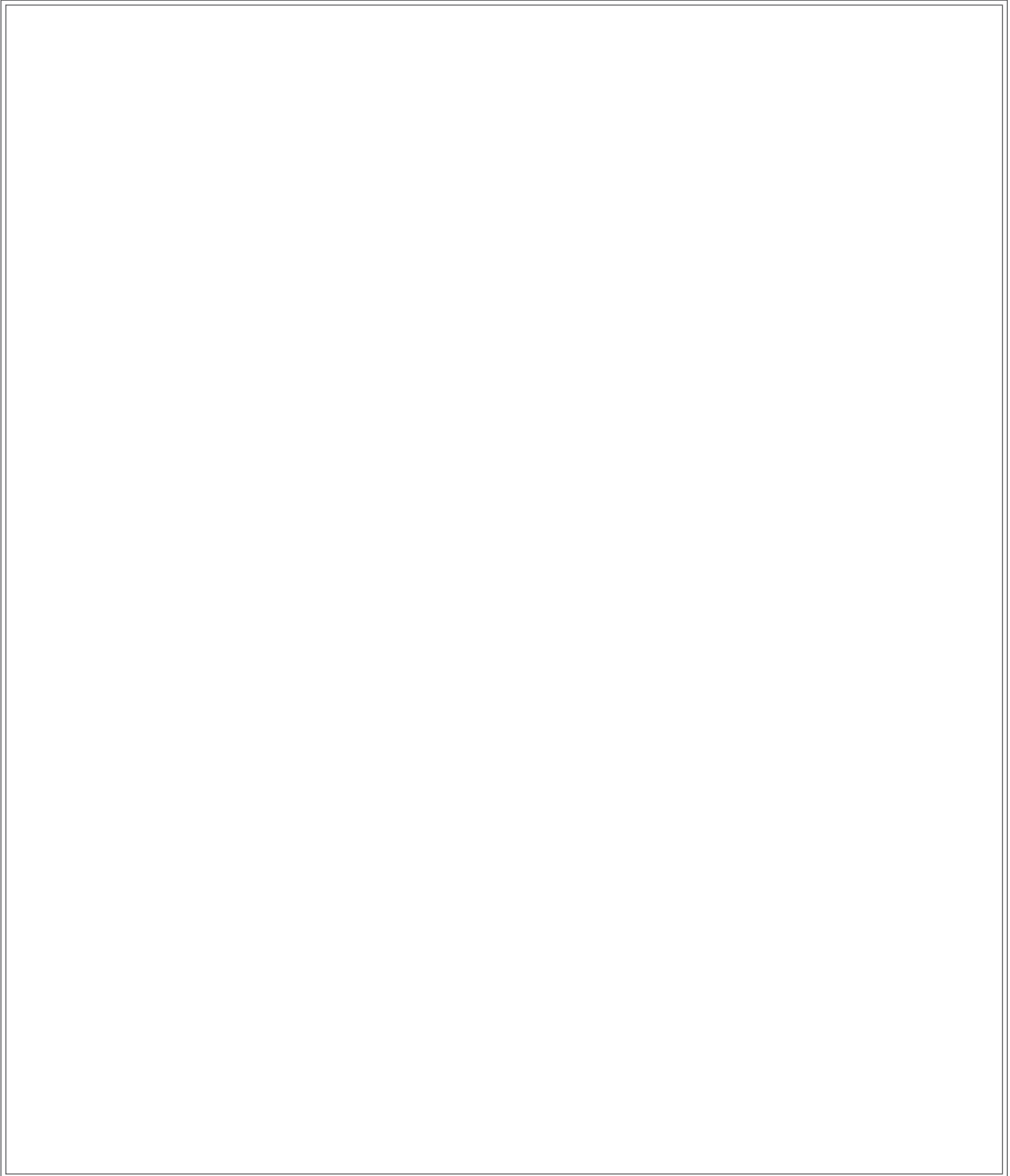Associate Professor of Biomedical Engineering,
Johns Hopkins University
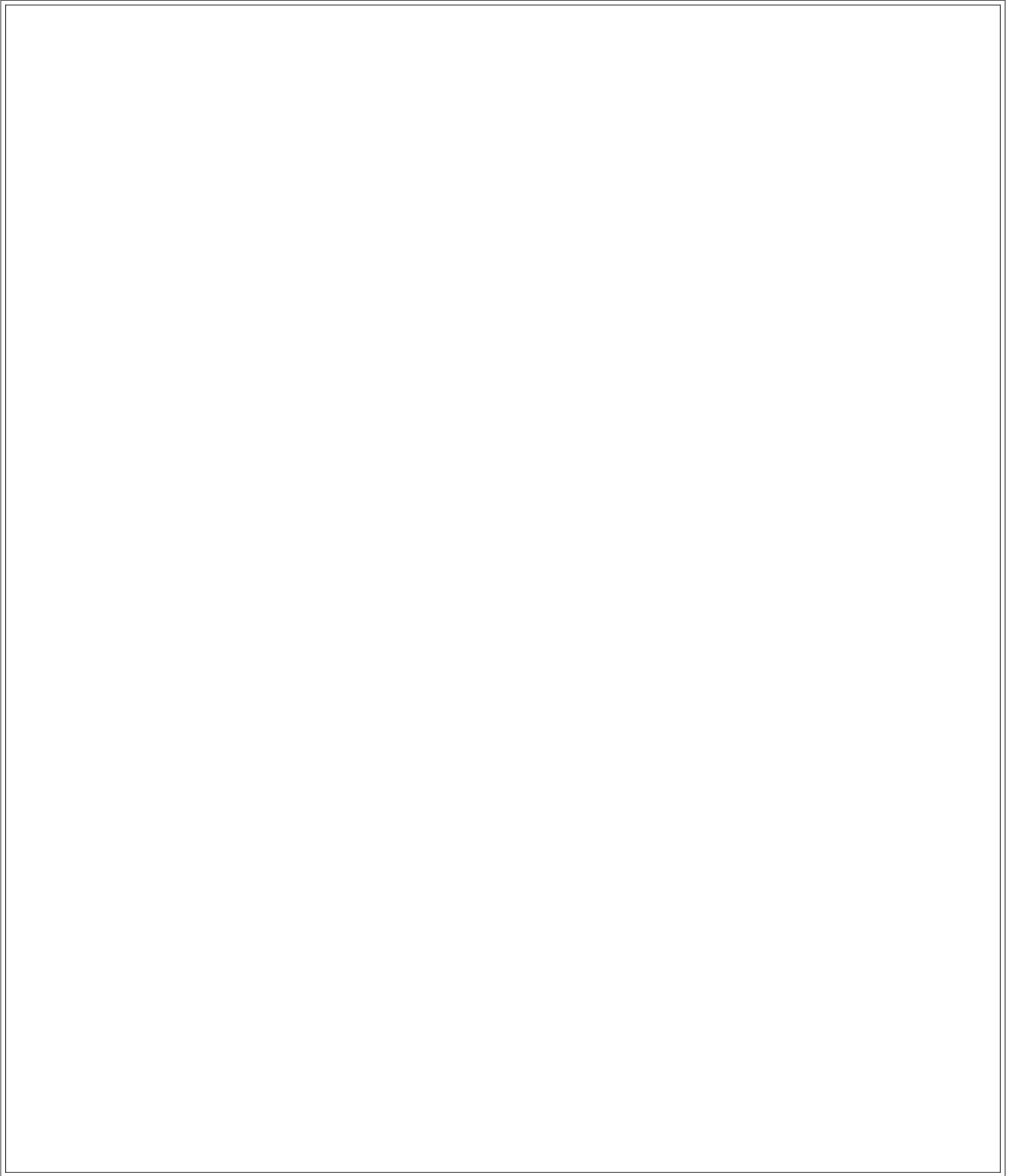
Yi Zhang, Ph.D.
Professor, Harvard Medical School

Notes

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Thanks to our Sponsors!

The Jackson Laboratory is an independent, nonprofit biomedical research institution with more than 2,200 employees. Headquartered in Bar Harbor, Maine, it has a National Cancer Institute-designated Cancer Center, a genomic medicine institute in Farmington, Conn., and facilities in Ellsworth, Maine, Sacramento, Calif., and Shanghai, China. Its mission is to discover precise genomic solutions for disease and empower the global biomedical community in the shared quest to improve human health. For more information, please visit www.jax.org.

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