Genetics, Genomics and Personalized Medicine Academic Plan Progress Report
Year 1 (2015-2016)

Enhance UConn's competitiveness in obtaining training grants such as the NSF Research Traineeship Program or the NIH Pre-doctoral Training in Biomedical Big Data Science.

- New training courses and workshops
  - 2 Bioinformatics workshops
  - 2 mRNA-seq workshops
  - 1mRNA workshop (summer 16)
  - 1 small RNA workshop (summer 16)
  - Data analysis and R workshops (summer 16)
  - minION training workshop (summer 16)
- The Center for Genome Innovation (CGI) has held several outreach workshops, including:
  - hosting participants in the Connecticut Junior Sciences & Humanities Symposium
  - hosting 8th grade class of the Charles H. Barrows STEM Academy
  - tour for Exploring Expertise pre-college experience for High School students and their parents
  - The Director of the CGI, Dr. Rachel O'Neill, was a recent guest speaker at the UConn Alumni College Association event held in Tampa
  - Academy of Biomedical Sciences, Martin Kellog Middle School tour and workshop
  - STEM Guidance Counselors tour
  - Guest Lecture and Tour for Diagnostic Genetic Sciences and Medical Laboratory Sciences students

- **MEDS 5325: Computational Genomics Practicum**
  The newly offered MEDS 5325 course focuses on methods for processing/analyzing NGS data that includes: introduction to the Linux command line, elements of Python and R programming; genomics software tools for performing sequence read-alignments, transcript-expression profiling, and robust procedures for gauging differential gene expression; methods for genome assembly, genome variation detection, motif-finding, and data-visualization, and statistical topics on probability distributions, central limit theorem, hypothesis testing, linear models, and dimensionality reduction.
  http://graveleylab.cam.uchc.edu/WebData/mduff/MEDS_5325_fall_2016.html
- **MEDS 6498: Machine Learning for Genomics**
  The MEDS 6498 course focuses on the elements of statistical learning, from classical methods for regression and classification to state-of-the-art deep learning architectures
  http://graveleylab.cam.uchc.edu/WebData/mduff/MEDS_6498_SPRING_2016/MEDS_6498_spring_2016.html
- Professional Science Master's Degree in Health Care Genetics:
  - 1st two graduates of the Program, May 2016
  - 3 new students start fall 2016 for total enrollment of 7
  - 2 students on internships in summer 2016 (Dartmouth Norris Cotton Cancer Center and Clinical Genomics and Advanced Technology at Dartmouth)

- 1st Next Generation Sequencing Module with a clinical case scenario taught over 3 days in the CGI by Dr. Bo Reese. Co-sponsored by the PSM in Health Care Genetics and Dr. Sal Frasca in Pathobiology

- **Successfully submitted genetic counselors proposal for 2nd phase of Academic Plan** entitled: "Genetics, Genomics & Counseling Program at the University of Connecticut". The goal of this initiative to establish a professional graduate level training program for genetic counselors which is important given the ever increasing number of genetic variants associated with human disease.
- **Awarded PAES funding** from the Provost's Office, Classroom Support Supplement Proposal – Institute for Systems Genomics/Engineering and Science Building space.
  - Funding supported the outfit of our teaching laboratory with USB-based, hand held DNA sequencers and mobile devices.
- One PhD student accepted into the ISG PhD program (self-funded). Due to financial constraints, admissions were suspended Fall 2015.

Build on successful expertise and extramurally funded projects that analyze big genome data sets and that develop cutting-edge genomics technology in order to increase our portfolio of consortium awards

- R21 entitled "High throughput validation of functional variants that affect pre-mRNA splicing. The goal of this project is to develop a massively parallel reporter assay to monitor the impact of genomic variants on pre-mRNA splicing.
- UConn-Wesleyan Stem Cell Core awarded. This award provides advanced genomics technologies in support of hESC production and editing, including: 1) Assessment of single cell gene expression; 2) Development of new genomic assessment tools; and 3) Implementation of global transcriptome-based approaches.
- Two NSF awards: 1) Standards and CyberInfrastructure That Enable "Big-Data" Driven Discovery for Tree Crop Research; and 2) Collaborative Research: ABI Development: Enabling Association Mapping for Tree Biology Research Through Advanced Integration of Genotype, Phenotype, and Geospatial Data.

Make UConn competitive for the ENCODE re-submission ($10M/4 years) as well as similar large awards in years 2 and 3.

- Encode grant was resubmitted.

Partner with the proposed Institute for Brain, Cognition and Behavior in, for example, correlating genotyping and neuroimaging with behavior and language disorders.

- Dr. Michael O'Neill, affiliate in Connecticut Institute for the Brain and Cognitive Sciences, was awarded IBACs seed grant and Graduate Fellowship.
- Dr. Marc Lalande gave a presentation at the Connecticut Institute for the Brain and Cognitive Sciences event on March 31, 2016.

Grow our genomics research and big data analysis capabilities at UConn to compete in these large federal and state funding opportunities.

Center for Genome Innovation:

- The CGI has expanded our capacity through the acquisition of several platforms that offer sequencing "long read" sequencing information: Oxford Nanopore MinION (in research and classroom training), Oxford Promethion, 10X Genomics GemCode – adding the following capabilities: simulated long-read sequencing, haplotype phasing, and single cell capture and library preparation (Illumina chemistry).
- The CGI has increased our throughput through the acquisition of an Illumina Neoprep, an automated liquid handler that will significantly reduce the time needed to produce Illumina libraries for DNA-Seq and mRNA-Seq, as well as sample input needed.
- We worked with the Office of the Vice President for Research to provide a subsidy to offset high-volume sequencing costs with external vendors.
- Metrics for the ISG reflecting growth in data analysis:
  - The Directors of the Biotechnology Bioservices Center (BBC), CGI, and the ISG have worked together to form the Computational Biology Core (CBC), a parallel unit to the CGI. Dr. Jill Wegrzyn as Director of this Core, which will continue to serve the computational biology needs of the university, agnostic to discipline, while expanding our capacity to support genomics-based data analyses.
  - Tendered two offer letters for Facility Scientist positions for the newly formed CBC.
  - Purchased equipment to expand our data analysis capacity, and have established a pipeline to merge this capacity with that available in the Center for Cell Analysis and Modeling Facility, under the direction of Dr. Ion Moraru.
- Metrics for the CGI reflecting growth in genomics research:
  - Illumina Hiseq Activity through CGI:
    - Two vendors: Macrogen and the New York Genome Center
    - Total lanes shipped through CGI: 103
    - Total billed through CGI: $212,340
  - Billable genomics services:
    - Through 12/31/2015: $72,800.52
    - 1/1/16 through 5/20/2016: $101,375.49
    - Total Billable Genomics Services FY16 to date: $303,461.76
    - Total PIs: 63 (35 Storrs, 2 Avery Point, 23 UConn Health, 3 External)
    - Departments at UConn Storrs: Animal Science (6), Molecular and Cell Biology (10), Nutritional Sciences (1), Pathobiology (2), Ecology and Evolutionary Biology (4), Physiology and Neurobiology (6), Nursing (2), Plant Sciences (1), Kinesiology (1), Dean's Office (1), Marine Sciences (2), Computer Science (1)
    - Departments/Centers at UConn Health: Vascular Biology (2), Community Medicine (1), Genetics and Genome Science (6), Reconstructive Science (2), Orthopaedic Surgery (3), Neuroscience (1), Psychiatry (1), Surgery (1), Neurology (1), Immunology (1), Medicine (1), Pediatrics (2), Neag Comprehensive Cancer Center (1)
    - External: Yale, Walter Reed Army Medical Center, Plum Island
- Vendor Tables, Product shows, open office hours and application seminars

- Letters of support and consulting activities:
  - Departments and UConn Storrs: Animal Science (7), Molecular and Cell Biology (7), Nutritional Sciences (1), Pathobiology (3), Ecology and Evolutionary Biology (3), Physiology and Neurobiology (3), Nursing (1), Plant Sciences (1), Kinesiology (2), Dean's Office (1), Marine Sciences (2), Computer Science (1)
  - Departments/Centers at UConn Health: Vascular Biology (2), Genetics and Genome Science (1), Reconstructive Science (1), Orthopaedic Surgery (3), Psychiatry (1), Surgery (1), Neurology (1)m Immunology (1) Medicine (3), Pediatrics (3), Neag Comprehensive Cancer Center (1), The Jackson Laboratory (1), Cell Biology (2), Dental Medicine (1), Orthodontics (1)
  - External: Yale, Walter Reed Army Medical Center, Plum Island, Shoreline Bio

Computational Biology Core

The directors of the BBC, CGI, and ISG worked together to form the Computational Biology Core (CBC), a parallel unit to the CGI, to support both UConn Health and the Storrs campus through the new director, Jill Wegrzyn, and part-time support members: Michael Duff (Assistant Professor) and Jeff Lary (System Administrator). In addition, an advisory committee has been formed to work in consultation with the CBC to serve both campuses, agnostic of discipline, while expanding the capacity for genomics-based data analyses. The current membership is detailed on the revised public website: http://bioinformatics.uconn.edu/people/. The CBC has completed interviews for Academic Assistants (staff Bioinformatic Scientists) and has extended offers to two candidates (funded jointly by the Academic Plan and the Office of the Vice President for Research). These individuals will take on the growing demand for bioinformatics support that exists on both campuses.

As provided through the Academic Plan, software purchases have been completed for commercial packages, including Geneious, CLC Genomics Workbench, and GeneXplain. The CBC is also collaborating with Dr. Daniel Schwartz, director of the new Proteomic Facility housed in the BBC, to select the new facility scientist and support their commercial software needs.

The expansion of the UConn Health's high performance computing cluster has been realized through equipment purchases funded by the Academic Plan. Efforts are underway to organize and document open-source packages on this cluster as a centralized resource. This is being conducted in collaboration with Dr. Ion Moraru and Stephen King at UConn Health. Jeff Lary will be assisting with the system administration needs as the user base on this system grows.

The facility has increased in volume in regards to analytical and hardware support to members of both campuses. Consulting activities include:

- Departments at UConn Storrs: Animal Science (7), Biomedical Engineering (1), Molecular and Cell Biology (8), Pathobiology (3), Ecology and Evolutionary Biology (9), Physiology and Neurobiology (3), Nursing (1), Pharmacy (3), Plant Sciences (5), Natural Resources (3), Marine Sciences (1), Statistics (2), Computer Sciences (3)

- Departments/Centers at UConn Health: Vascular Biology (2), Genetics and Genome Sciences (3), Orthopaedic Surgery (2), Psychiatry (3), Surgery (1), Neag Comprehensive Cancer Center (1), Cell Biology (2), Orthodontics (1)
- External: Plum Island (1)

We recently started to track grant awards resulting from our consulting/training efforts, totaling $4,484,785.

Additional information:

- Institute for Systems Genomics newsletters
  - September 2015
  - April 2016
- Institute for Systems Genomics workshops
  - Genomics, Family and Health Event with Rebecca Skloots and members of the Lacks Family
  - Genomics, Bioinformatics, and Gene Editing Workshop
  - Genomics Academic Plan and Technology Workshop
  - Institute for Systems Genomics Networking Workshop