Genomic Analysts at (or with affiliations to) CCHMC (presenters from the Seminar "Bridging the Gap" have an *)

Data Types

DNA: Variant (Post processed genotype data, raw genotype array data, raw sequence data, post processed sequence data) and methylation (chip based, pyrosequencing, bisulfite conversion sequencing). DNA is not tissue dependent but methylation is.

RNA: Gene Expression (Array, sequence), MicroRNA (miRNA), ChIP-Seq. RNA is tissue dependent. And be done in single cell.

Approaches

Isolated Effects: The purpose of identifying isolated effects (variant differences or single gene effect) is to identify biomarkers for disease or differential treatment. Single gene effects have been implicated in Mendelian diseases such as breast cancer and Huntington's disease. Identification of specific variants or gene dysregulation may help predict prognosis or optimal treatment strategy.

Effects of Variants: The purpose of identifying effects of variants is to predict whether a variant would be expected to have a biologic effect. These effects could be due to changes in protein structure, changes in splice sites, changes in transcription factor binding sites. This work could justify further functional studies.

Biologic Networks: The purpose of identifying biologic networks is to identify gene signaling pathways which are altered with a state (disease, treatment status). These signaling pathways may provide important insight into the specific changes that would be missed by looking at a single gene. Ultimately these approaches will identify novel therapeutic targets and may allow the avoidance of side effects. In addition by understanding the pathways which have altered regulation we have better understanding of the underlying biologic changes.

Complex interactions of the genome: The purpose of identifying complex interactions of the genome is to understand the big picture of how all of the pieces interact (in an organism, tissue, or cell). These could include interaction within a single organism's genome or between genomes (human genome with the gut microbiome).

Primary Approach

Isolated Effects

***Ge Zhang, MD, PhD** (ge.zhang@cchmc.org)
Associate Professor, Human Genetics
Focus: Explaining variation and evolutionary influences of complex traits (cardiometabolic traits, birth timing)
Data Types: DNA (genotype and sequence)
Secondary Approach: Multi-gene effects
Methods Development: Variant calling from next-generation sequencing data, storage and processing of genomic data

***Leah Kottyan, PhD** (leah.kottyan@cchmc.org)
Assistant Professor, CAGE
Focus: Gene identification
Data Types: DNA (genotype and sequence), RNA (RNA-seq)

Lisa Martin, PhD (lisa.martin@cchmc.org)
Professor, Human Genetics
Data Types: DNA (sequence and genotype), methylation
Focus: Integration of statistical genetics with biology and epidemiology to understand disease and variation (heart malformations, asthma, eosinophilic esophagitis, drug response)
Secondary Approach: Gene networks, multi-gene effects
Methods Development: Variant discovery strategies (VIOLET) for family based data, Quality control pipelines for DNA variants

Valentina Pilipenko, PhD (valentina.pilepenko@cchmc.org)
Biostatistician I, Human Genetics
Data Types: DNA (genotype and sequence)
Methods Development: Quality control pipelines for DNA variants

Xue Zhang, PhD (zue.zhang@cchmc.org)
Biostatistician II, Human Genetics
Data Types: DNA (genotype), methylation
Secondary Approach: Regional effects

Hua He, MS (hua.he@cchmc.org)
Biostatistician II, Human Genetics
Data Types: DNA (genotype), RNA (array based)
Secondary Approach: Gene Networks

Ken Kaufman, PhD (Kenneth.Kaufman@cchmc.org)
Professor, CAGE
Focus: Gene Identificaiton
Data Types: DNA (genotype and sequence)

Bahram Namjou-Khales, MD (Bahram.Namjou@cchmc.org)
Assistant Professor, CAGE
Focus: Characterization of subphenotypes in genetic analyses (lupus, EMR based data)
Data Types: DNA (genotype)
Methods Development: Algorithm development to establish phenotype from the electronic medical record.

Kristen Sund, PhD (Kristen.Sund@cchmc.org)
Project Manager, Cincinnati Children's Research Foundation
Focus: Identification of clinically actionable variants.
Data Types: DNA (sequence)

Rob Hufnagel, MD, PhD (Robert.Hufnagel@cchmc.org)
Resident, Human Genetics
Focus: Identification of clinically actionable variants (strong clinical interest in eye, but anything genetic is of interest)
Data Types: DNA (sequence)

Nicole Weaver, MD (Kathryn.Weaver@cchmc.org)
Instructor, Human Genetics
Focus: Identification of clinically actionable variants (clinical interest in craniofacial malformations, heart)
Data Types: DNA (sequence)

Lili Ding, PhD (Lili.Ding@cchmc.org)
Assistant Professor, Biostatistics and Epidemiology
Focus: To utilize advanced mathematical statistical approaches to help account for the complex inheritance of common traits.
Data Types: DNA

Tesfaye Mersha, PhD (Tesfaye.Mersha@cchmc.org)
Assistant Professor, Asthma Research
Focus: Ancestry mapping to localize asthma liability genes in admixed population; unravel genetic from non-genetic factors in the etiology of asthma using population isolates.
Data Types: DNA, RNA, methylation
Secondary Approaches: Gene expression, proteomics,
Methods Development: *AncestrySNPminer*, the first Web-based bioinformatics tool specifically designed to retrieve genome-wide Ancestry Informative Markers (AIMs) from public or private databases (HapMap, 1000 Genomes Project, etc.)

John Harley, MD, PhD (John.Harley@cchmc.org)
Director, CAGE
Focus: Understanding of the genesis of lupus and other immunological diseases
Data Types: DNA (genotyping arrays and sequencing)
Peter White, PhD (Peter.White@cchmc.org)
Director, Biomedical Informatics
Focus: Genomic contributors to disease risk for pediatric disorders, including ADHD, cardiac defects, and solid tumors
Data Types: DNA (sequencing)
Secondary Approaches: Variant effect, incorporating phenotypic information, bioinformatics tools for data integration
Methods Development: New methods for extracting integrating and analyzing biomedical data using novel informatics approaches and innovative approaches for integrating and disseminating clinical, phenotypic, and molecular data to researchers for promoting discovery and hypothesis validation

Effects of Variants

***Artem Barski, PhD,** (Artem.Barski@cchmc.org)
Assistant Professor, Allergy and Immunology
Focus: epigenetic and transcriptional regulation of gene expression
Data Types: RNA, CHiP-seq, epigenomics
Secondary Approach: Epigenenomics
Methods Development: computational solutions to the high level data being generated including an epigenomics data analysis system (Wardrobe)

Matt Weirauch, PhD (Matthew.Weirauch@cchmc.org)
Assistant Professor, CAGE
Focus: Development of models of gene regulation in both normal and disease states
Identification of Variants which Influence: Transcription factor binding and RNA binding
Methods Development: Development of tools to identify transcription factors and RNA binding proteins

Alexey Porollo, PhD (Alexey.porollo@cchmc.org)
Assistant Professor, CAGE
Focus: Protein structure, protein-protein interaction

Nan Shen, MD (Nan.Shen@cchmc.org)
Associate Professor, CAGE
Focus: Impact of miRNA, lncRNA

Jarek Meller, PhD (JMeller@cchmc.org)
Associate Professor, Biomedical Informatics
Focus: Computational approaches for data mining, analysis and knowledge extraction from biomedical data
Methods Development: Tools for functional and structural annotation of proteins and their complexes (SABLE, SPPIDER and POLYVIEW-3D)

Biologic Networks

***Yan Xu, PhD** (Yan.Xu@cchmc.org)
Associate Professor, Neonatology and Pulmonary Biology
Focus: Gene networks for lung development and disease.
Data Types: mRNA, miRNA array, seq, ChIP seq, Single Cell Transcriptome.
Methods development: Need based analytic pipelines

Anil Jegga, DVM, MRes (Anil.Jegga@cchmc.org)
Associate Professor, Biomedical Informatics
Focus: Comparative and functional genomics (transcriptional and posttranscriptional regulatory networks, biological network analyses, phenotypic networks and systems biology of disease)
Data Types: RNA (miRNA, array), DNA
Secondary Approaches: Effect of variants
Methods Development: Development of a one-stop portal for gene list enrichment analysis and candidate gene prioritization (ToppGene Suite) and a tool that enables discovery of shared cis-elements in conserved non-coding sequences of mice and humans (GenomeTraFaC)

Ping-I (Daniel) Lin, MD, PhD (Ping-I.Lin@cchmc.org)
Assistant Professor, Biostatistics and Epidemiology
Focus: Studying the genomics in neuropsychiatric conditions, particularly endophenotypes. Applying computational algorithms (e.g., genetic algorithm) to the multi-modal data consisted of genomic, neurochemical, neuroanatomic, and behavioral data of "constructs" relevant to neuropsychiatric disorders.
Data Types: DNA, RNA
Secondary Approaches: Interactions of the genome, variant/gene effects, and effects of variants

Nathan Salomonis, PhD (Nathan.Salomonis@cchmc.org)
Assistant Professor, Biomedical Informatics
Focus: Examine the interplay between diverse modes of gene regulation (splicing, transcription) in human disease and development
Data Types: RNA (including single cell approaches)
Secondary Approaches: Pathway, network (genes and isoforms)
Methods Development: Open access tools (AltAnalyze) for gene expression and splicing analysis (http://altanalyze.org). These tools are designed for non-informatics biologists.

Jun Ma, PhD (Jun.Ma@cchmc.org)
Professor, Biomedical Informatics
Focus: Molecular mechanisms of gene regulation and embryonic development
Data Types: RNA

Mario Medvedovic, PhD (Mario.Medvedovic@uc.edu)
Associate Professor, UC Environmental Health
Focus: Developing and applying new statistical and computational procedures for the analysis of complex genomic, functional genomic data
Data: RNA, DNA, methylation, proteomics

Rebekah Karns, PhD (Rebekah.Karns@cchmc.org)
Research Fellow, Biomedical Informatics
Focus: Genomic Analyst for the Digestive Health Center
Data: RNA, DNA
Secondary Approaches: Isolated effect

Mihaela Pavlicev, PhD (mihaela.pavlicev@cchmc.org)
Assistant Professor, Neonatology and Pulmonary Biology
Focus: Comparative Genomics
Data Types: DNA, RNA

Steve Potter, PhD (steve.potter@cchmc.org)
Professor, Developmental Biology
Focus: Understanding the principle of organogenesis by defining gene-expression programs driving development
Data Types: RNA, DNA, single cell gene expression profiles

Complex Interactions of the Genome

***Long (Jason) Lu, PhD** (Long.Lu@cchmc.org)
Associate Professor, Biomedical Informatics
Focus: Bringing quantitative approaches from disciplines such as computer science and applied mathematics to study the molecular mechanism of human diseases.
Data Types: Transcriptome and whole genome data from the metagenome (additionally) Exome Seq, CHiP-Seq)
Secondary Approaches: Isolated Effect, Gene Networks

*** Kakajan Komurov, PhD** (Kakajan.Komurov@cchmc.org)
Assistant Professor, Hematology and Oncology
Focus: Building global mechanistic models of cancer progression.
Methods Development: Novel algorithms for extraction of network models from multidimensional genome scale data, development a platform for network analysis in functional genomics (NetWalker).

Bruce Aronow, PhD (Bruce.Aronow@cchmc.org)
Professor, Biomedical Informatics
Focus: Characterizing biological systems as dynamic networks that relate the structure, function, and expression of genes, transcripts and proteins in cellular, physiological, and disease-affected context
Secondary Approaches: gene networks, effect of variant, isolated effects,
Data Types: RNA (array, RNA-seq, ChIP-seq), DNA (sequence, array)
Methods Development: Development of a one-stop portal for gene list enrichment analysis and candidate gene prioritization (ToppGene), and a tool that enables discovery of shared cis-elements in conserved non-coding sequences of mice and humans (GenomeTraFaC)

***Patrick Putnam** (Patrick.Putnam@cchmc.org)
Application Developer, Human Genetics
Focus: Addressing scalability, optimization, and parallelization of scientific problems
Data Types: DNA, RNA
Secondary Approaches: isolated effects
Methods Development: Computational approaches to big data.

Eileen Alexander, PhD (Eileen.Alexander@cchmc.org)
Assistant Professor, Xavier University
Focus: Complex interactions of the genome, such as heritability analyses in families and twins to determine the relative effects of genetics and environmental factors
Data Types: methylation, DNA variants, gene expression
Secondary Approaches: Dynamic changes in expression, as they relate to epigenomic alterations

Nupur Dasgupta, PhD (Nupur.Dagupta@cchmc.org)
Research Associate, Division of Human Genetics
Focus: Characterizing networks, functions, pathways, transcription regulators associated with different physiological conditions or drug treatments in disease models
Data Types: mRNASeq , miRNASeq, mRNA array, miRNA array, miRNA-mRNA interactions
Secondary Approaches: Identifying differentially expressed genes and transcripts, comparison of RNA-seq analytical methods, comparing RNA SEQ and Array data

Genomic Analysts at (or with affiliations to) CCHMC (presenters from the Seminar "Bridging the Gap" have an *)

Iouri Chepelev, PhD (Iouri.Chepelev@cchmc.org)
Assistant Professor, CAGE
Focus: Chromatin Structure

Hector Wong, MD (hector.wong@cchmc.org)
Professor of Pediatrics, Critical Care Medicine
Focus: Translating transcriptome data for biomarker discovery and development, and for the identification of disease subclasses
Data Types: RNA

Individuals Who Play an Essential Role but Don't Fall into the Categories

Michael Wagner, PhD (Michael.Wagner@cchmc.org)
Associate Professor, Biomedical Informatics
Focus: Large-scale optimization and applications in biomedical informatics
Role: As faculty liaison to the Biomedical Informatics Core, Dr. Wagner can help facilitate bioinformatics support
Methods Development: Developed rapid data flow infrastructure that includes parallelized genotype calling algorithms

Phillip Dexheimer, MS (Phillip.Dexheimer@cchmc.org)
Senior System Programmer, Biomedical Informatics
Focus: Processing of next generation sequence data
Types of Data: DNA, RNA