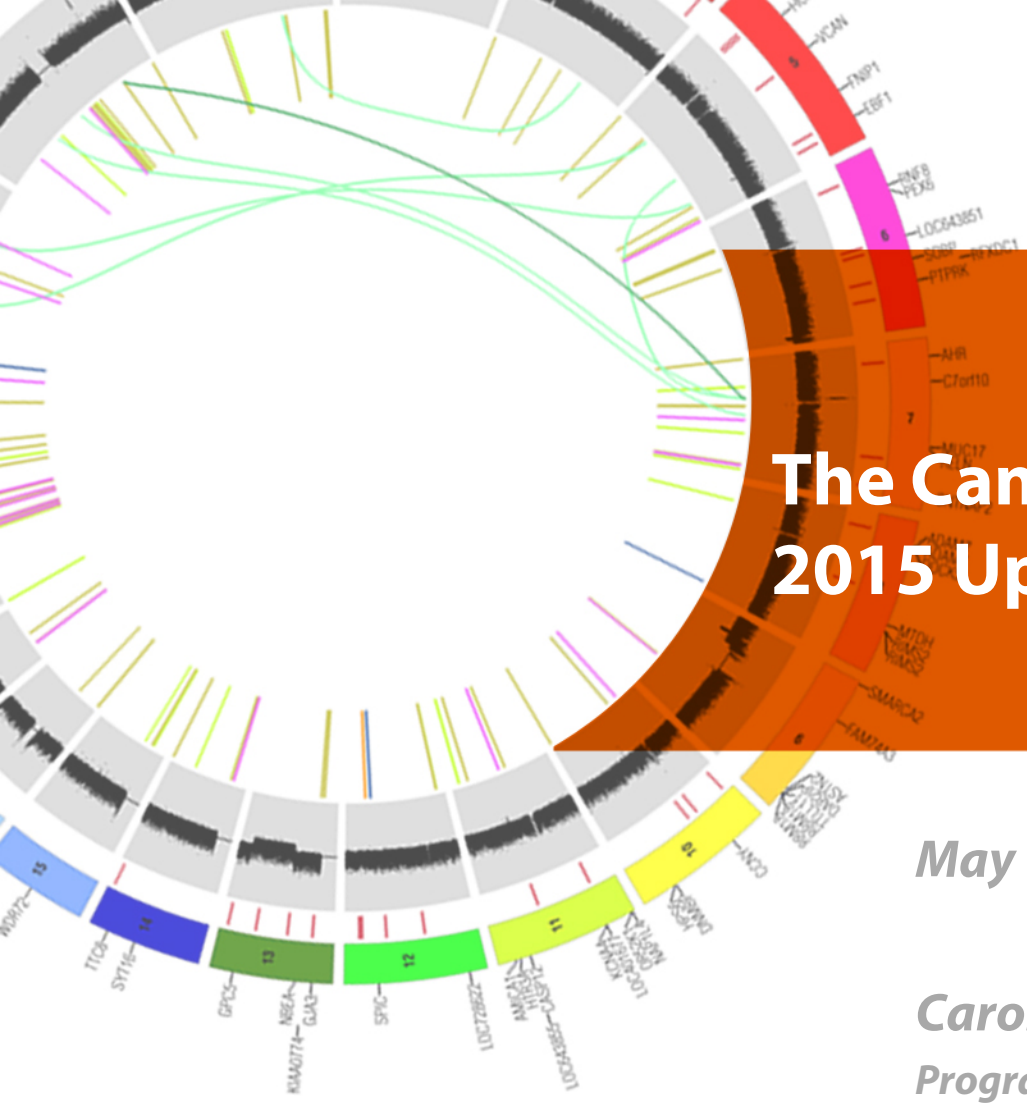
TCGA Team Named as 2015 “Sammies” Finalists

The Cancer Genome Atlas Team:
Mapped thousands of gene sequences for more than thirty types of cancer, advancing precision medicine in the diagnosis, treatment and prevention of these deadly diseases.

Samuel J. Heyman
SERVICE to AMERICA MEDALS



The Cancer Genome Atlas

The Cancer Genome Atlas: 2015 Update Part 2

May 11, 2015

Carolyn M. Hutter, Ph.D.

Program Director

Division of Genomic Medicine

National Human Genome Research Institute

Whole Exome Sequencing

- Sequence production completed.

- Multi-center mutation calling multi-tumor completion (MC3) effort.
  - Goal: Generate a high quality mutation call set for TCGA cases
  - Builds on lessons learned through TCGA multi-center mutation calling (MC2) exercises
  - Standardized calls across tumor types
  - “Containerizing” established mutation calling tools
  - Will facilitate Pan-Cancer analysis

- Continued tumor specific validation efforts to support AWG marker papers.

Whole Genome Sequencing

- WGS for 24 tumor types
  - 14 tumor types with >40 cases
  - Last cases to be uploaded in June (Illumina X Ten)

- Most samples being analyzed in ICGC/TCGA Pan-Cancer Analysis of Whole Genomes (PCAWG)

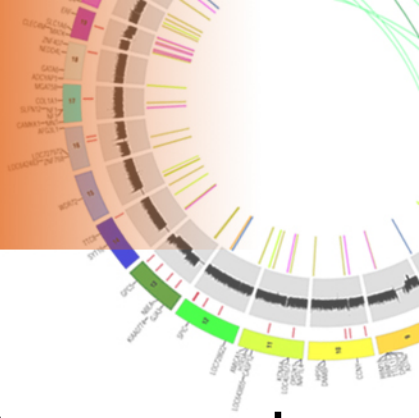
AACR American Association for Cancer Research ANNUAL MEETING 2015 | PHILADELPHIA
APRIL 18-22 | #AACR15 PENNSYLVANIA CONVENTION CENTER
Bringing Cancer Discoveries to Patients

> Return to AACR Annual Meeting

Session Title: Pan-Cancer Analysis of Whole Genomes
Session Type: Major Symposium
Session Start/End Time: Sunday, Apr 19, 2015, 1:00 PM - 3:00 PM
Location: Terrace Ballroom II-III (400 Level), Pennsylvania Convention Center
CME: CME-Designated
CME/CE Hours: 2
Session Description: The Pan-Cancer analysis of Whole Genomes (PCAWG) project of the International Cancer Genome Consortium (ICGC) and The Cancer Genome Atlas (TCGA) is co-ordinating analysis of more than 2,000 whole cancer genomes. Each genome is characterized through a suite of centralized algorithms, including alignment to the reference genome, standardized quality assessment and calling of all classes of somatic mutation. Scientists participating in the research projects of PCAWG are addressing a series of fundamental questions about cancer biology and evolution based on these data, a sample of which will be presented at this session. Key areas of study include: (1) Discovery of driver mutations outside of the protein-coding regions of the genome; (2) Integrating mutational signatures across tumor types and mutation categories; (3) Characterizing subclonal structures and patterns of genome evolution across cancers; (4) Investigating relationships between germline and somatic mutations; (5) Investigating biological pathways targeted by driver mutations.

Cancer genome analysis in the cloud: Technical, ethical and legal challenges
Sunday, Apr 19, 2015, 1:10 PM - 1:30 PM
Lincoln Stein. Ontario Institute for Cancer Research, Toronto, ON, Canada
Pathways and Drivers in 2,000 cancer genomes
Sunday, Apr 19, 2015, 2:00 PM - 2:20 PM
Joshua M. Stuart. UC Santa Cruz, Santa Cruz, CA
Investigation of germline genetic variation in 2,500 whole cancer genomes
Sunday, Apr 19, 2015, 1:35 PM - 1:55 PM
Jan Korbel. European Molecular Biology Laboratory, Heidelberg, Germany
Structural variation in 2,000 cancer genomes
Sunday, Apr 19, 2015, 2:25 PM - 2:45 PM
Peter J. Campbell. Wellcome Trust Sanger Inst., Cambridge, United Kingdom

Future Programs



- NHGRI large-scale sequencing efforts with increased focus on Mendelian and common disease
- Applications of cancer genomics in genomic medicine
- Precision Medicine Initiative (pending appropriation)

UP FOR A CHALLENGE? (U4C)

STIMULATING INNOVATION in BREAST CANCER GENETIC EPIDEMIOLOGY

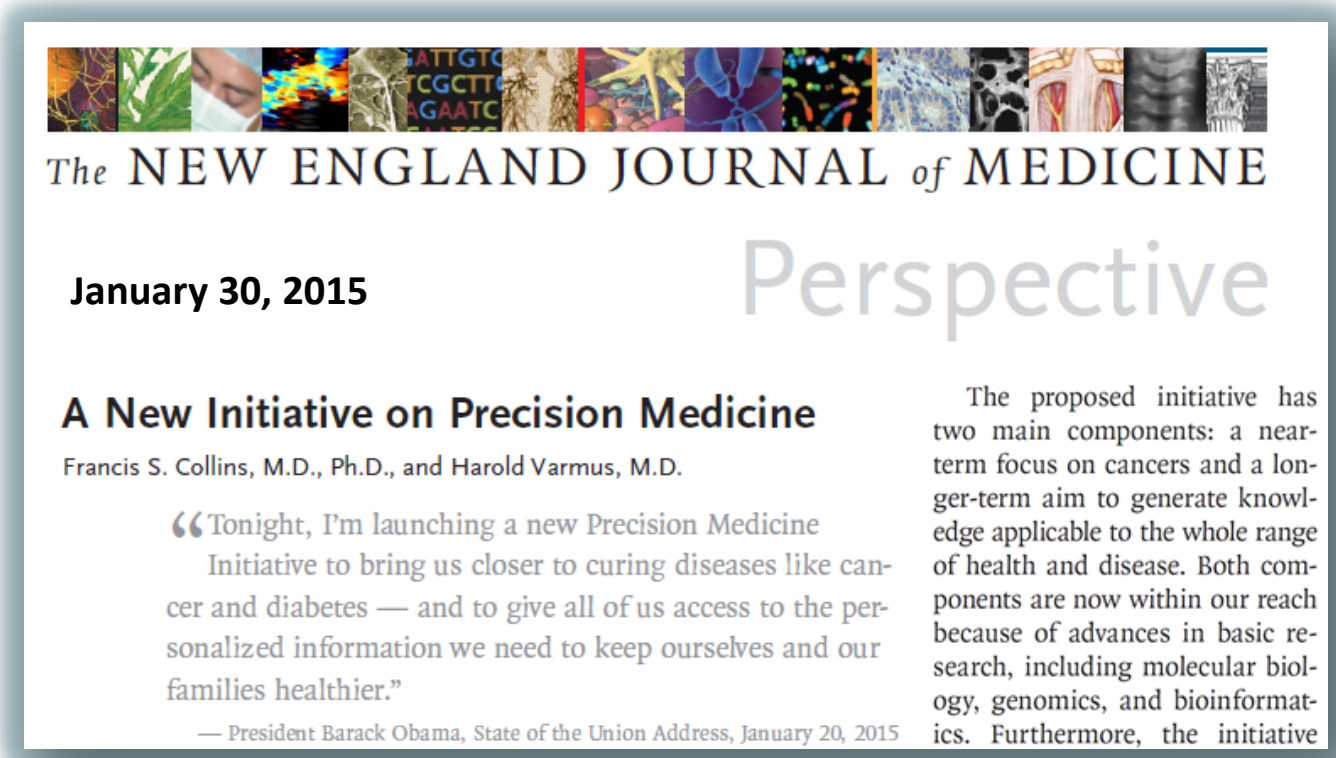
Coming Soon -- A Challenge Sponsored by the NCI

- **The Challenge: To use innovative approaches to identify novel pathways—including new genes or combinations of genes, genetic variants, or sets of genomic features—involved in breast cancer susceptibility**
- **Goals:**
  - **Provide further insights into genetic contribution to breast cancer**
  - **Advance innovation in breast cancer genetic epidemiology research**
  - **Increase amount and diversity of minds tackling the problem**
  - **Making breast cancer genetic data more widely available**
- **For more info: https://www.synapse.org/upforachallenge**

“And that’s why the budget I send this Congress on Monday will include a new Precision Medicine Initiative that brings America closer to curing diseases like cancer and diabetes, and gives all of us access, potentially, to the personalized information that we need to keep ourselves and our families healthier.”

President Barack Obama

January 30, 2015



The NEW ENGLAND JOURNAL of MEDICINE

January 30, 2015

Perspective

A New Initiative on Precision Medicine

Francis S. Collins, M.D., Ph.D., and Harold Varmus, M.D.

"Tonight, I'm launching a new Precision Medicine Initiative to bring us closer to curing diseases like cancer and diabetes — and to give all of us access to the personalized information we need to keep ourselves and our families healthier."

— President Barack Obama, State of the Union Address, January 20, 2015

The proposed initiative has two main components: a near-term focus on cancers and a longer-term aim to generate knowledge applicable to the whole range of health and disease. Both components are now within our reach because of advances in basic research, including molecular biology, genomics, and bioinformatics. Furthermore, the initiative

http://www.nih.gov/precisionmedicine/