BROAD INSTITUTE

iSeqTools
http://iseqtools.org/

GATK Workshop

University of Pretoria
South Africa, 22-26 June, 2015

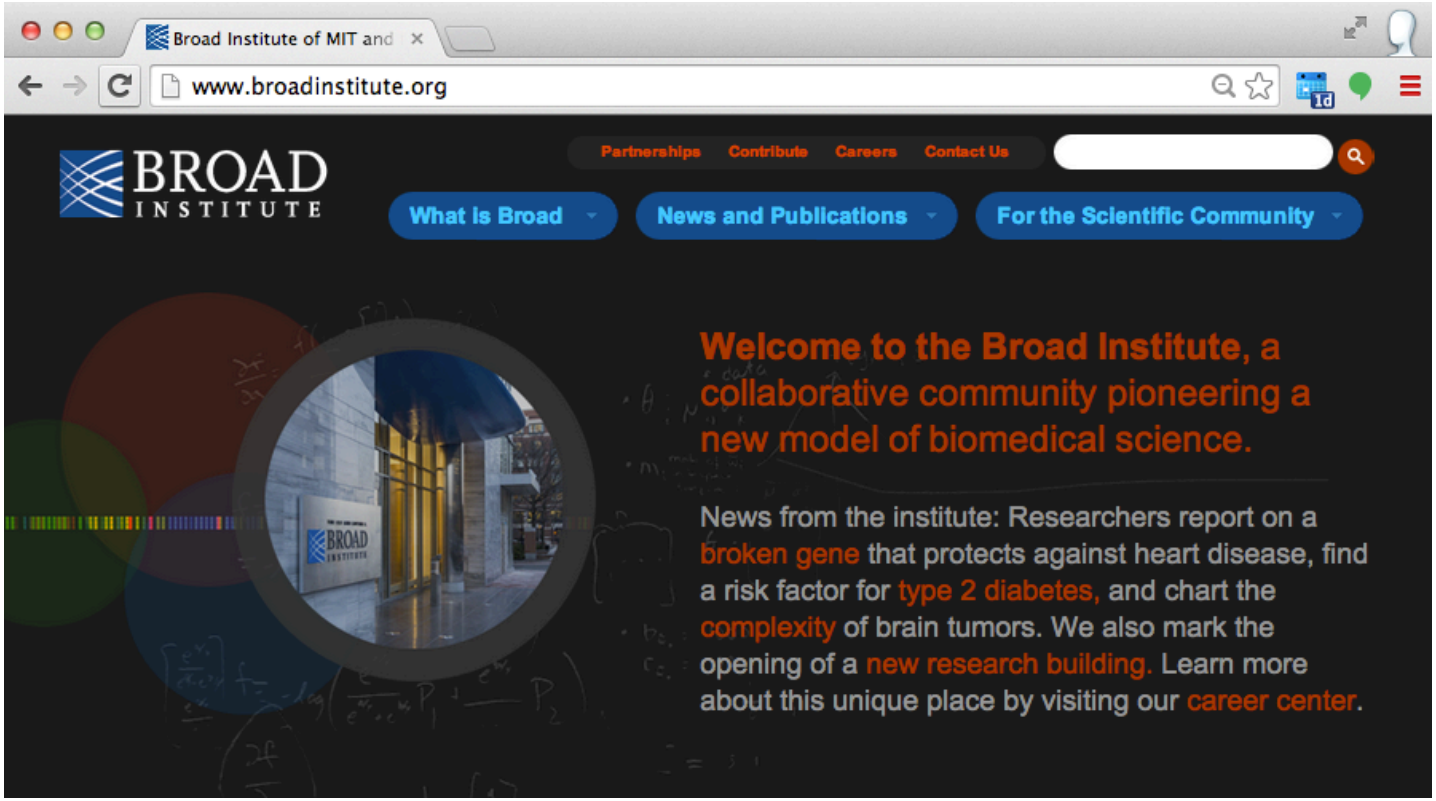
Data Sciences & Data Engineering
Broad Institute of Harvard and MIT
http://www.broadinstitute.org/gatk/

@gatk_dev

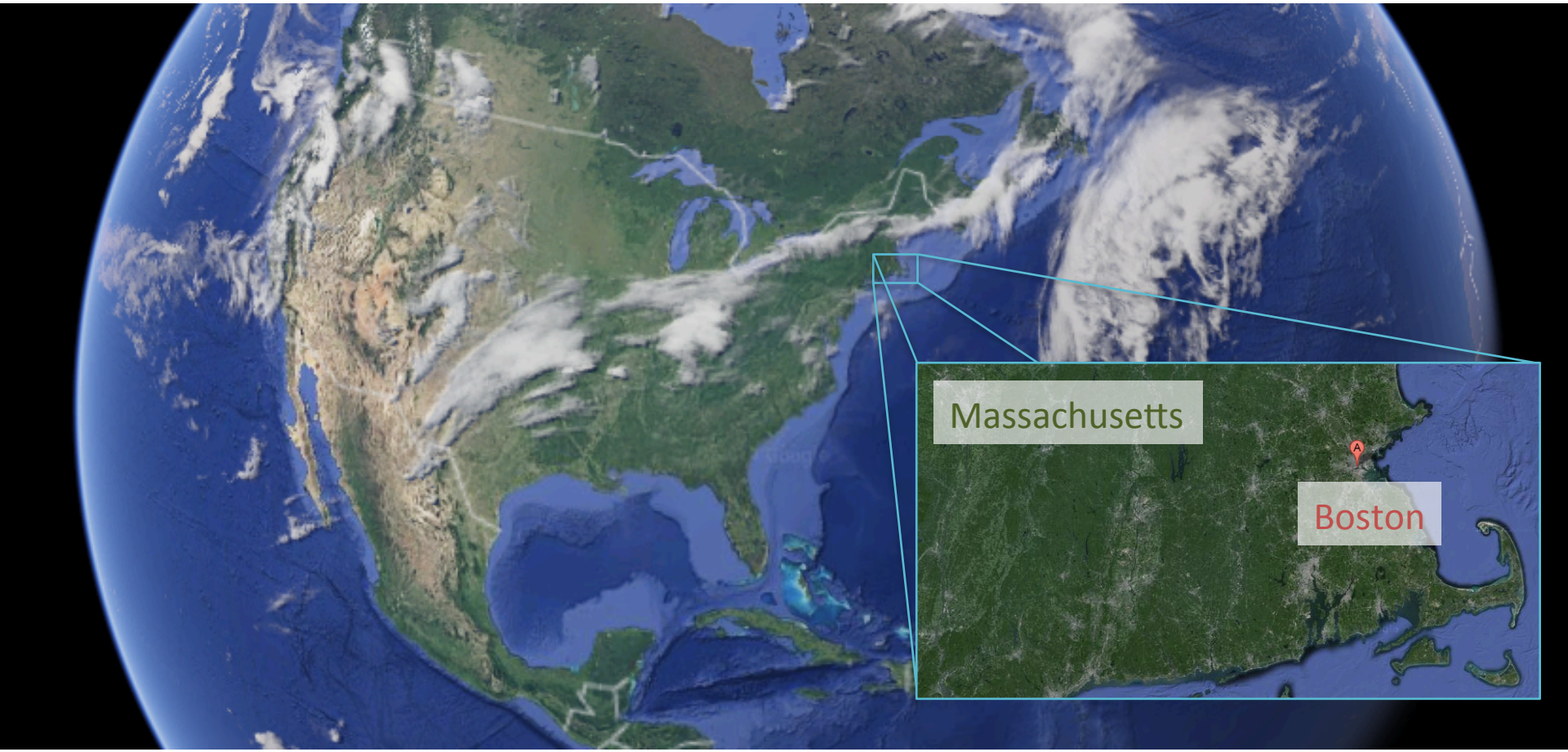
BROAD INSTITUTE

What / who is the Broad Institute?

- Spinoff of Harvard & MIT -- Eric Lander and philanthropists Eli & Edyth Broad
- Use the full power of genomics to transform the understanding and treatment of disease



Where in the world is the Broad?



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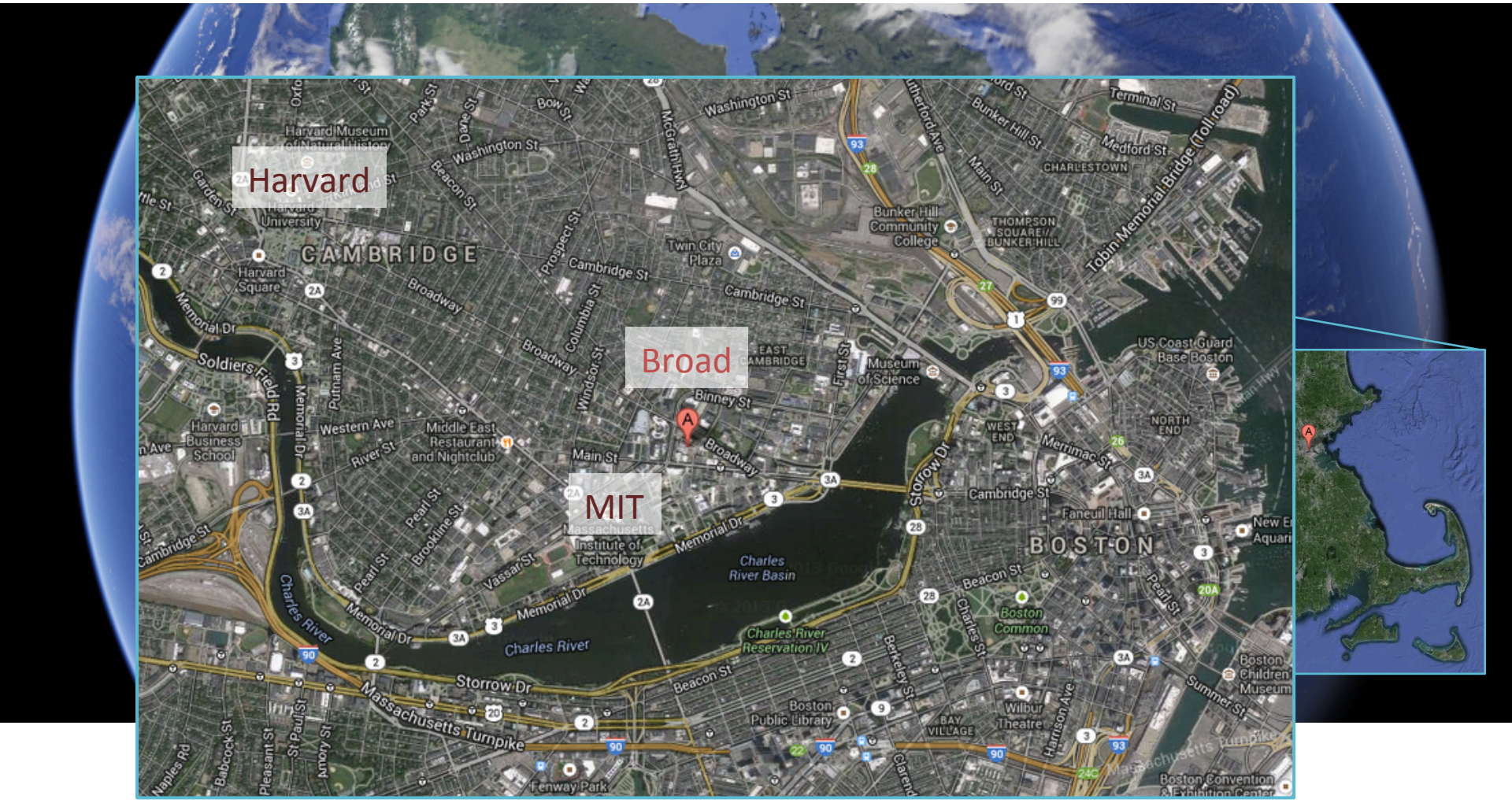

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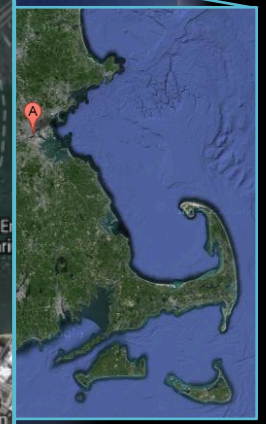
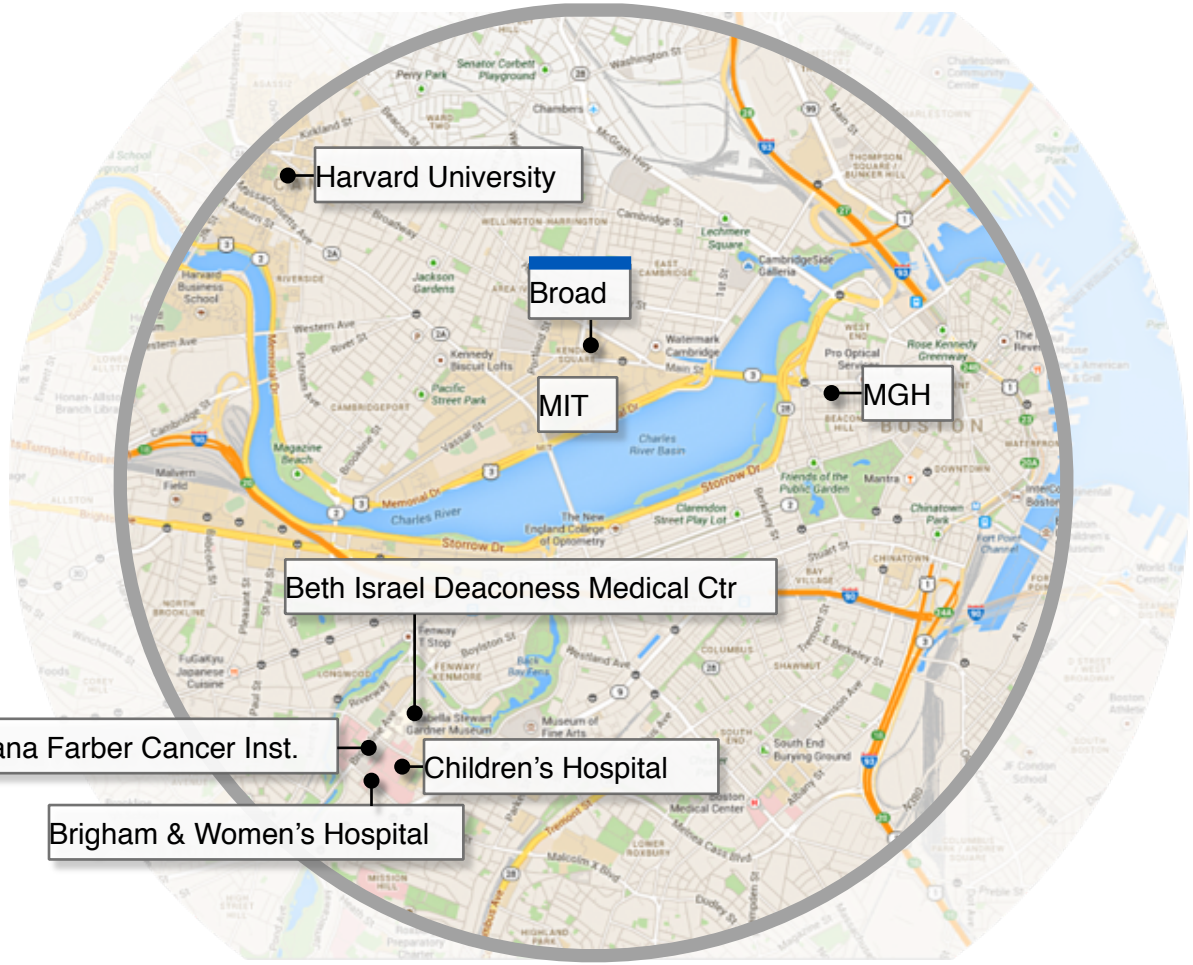
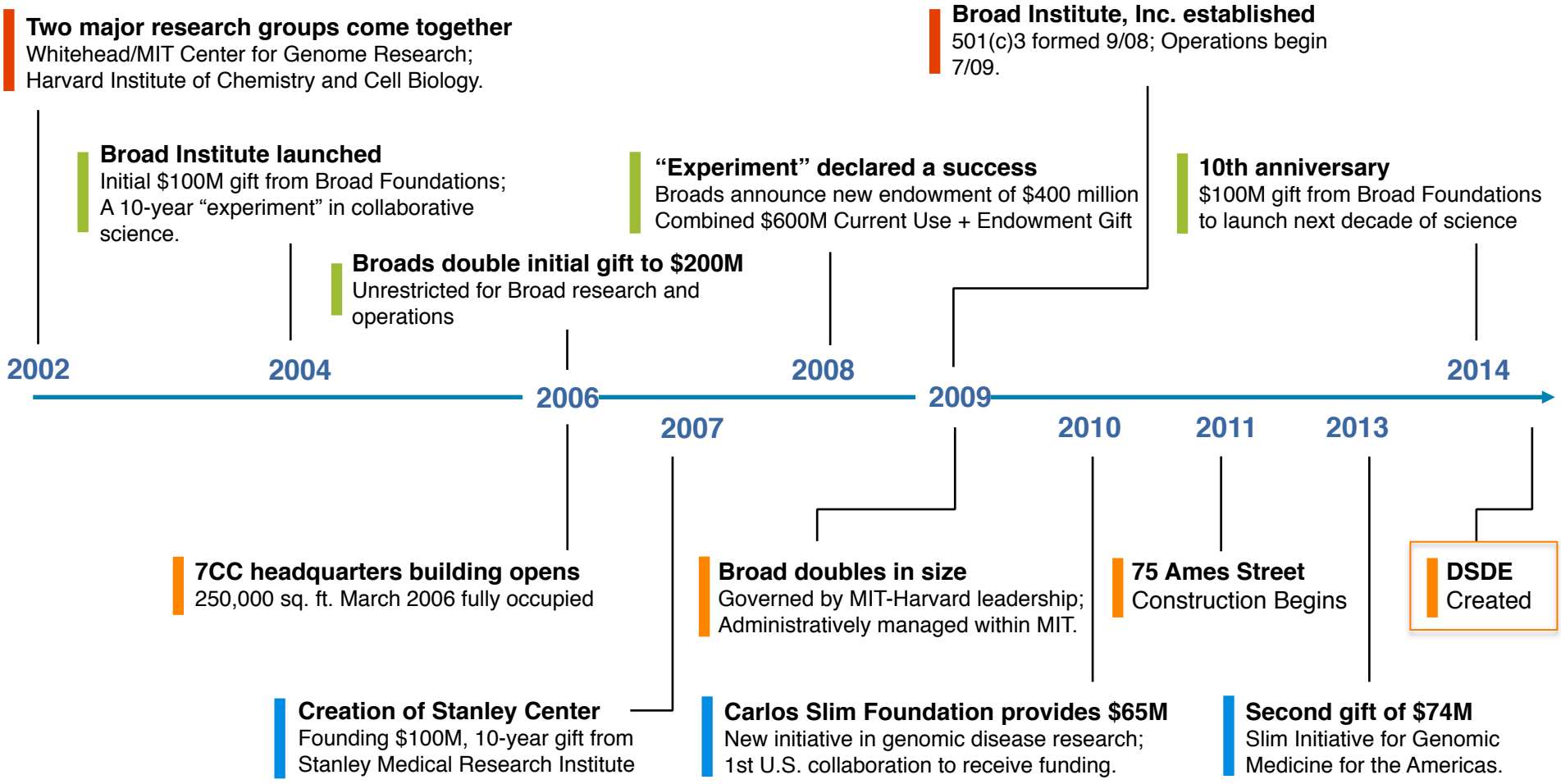


Where in the world is the Broad?

A highly collaborative and research-dense environment



A brief history of the Broad Institute



Data Science & Data Engineering @ Broad

A new organization bringing together software engineers, computational biologists, and computing infrastructure specialists.

A vision that articulates an advanced computing infrastructure, set of data and analysis services leveraging modern cloud computing paradigms.

https://www.broadinstitute.org/dsde/

DSDE's extended family: GP + CGA + BITS

DSDE: Methods + Engineering + Operations + Communications

Genomics Platform (GP)

Cancer Genome Analysis Group (CGA)

Broad IT (BITS)

+ many collaborators who beta-test the methods and software

General program

Day 1
Introductions, pipelines and QC

Day 2
Data pre-processing (BAM cleanup)

Day 3
Germline variant discovery

Day 4
Somatic variant discovery

Day 5
Working with variants

AM Lecture session: 9am – 12pm

PM Hands-on tutorial: 1pm – 4pm

Day 1: Sequencing for variant analysis

- 9:00 Workshop introduction
- 9:20 High-throughput sequencing for variant analysis
- 10:00 GATK Best Practices and the Broad pipelines
- *10:30 Coffee break*
- 11:00 Sequence data quality control
- 11:30 Pipelining and parallelism
- *12:00 Lunch break*
- 13:00 Hands-on session

Approximate schedule – timing may vary depending on Q&A

Day 2: Mapping and cleanup

- 9:00 Recap of Day 1 and Q&A
- 9:15 Mapping and pre-processing
- 10:00 Indel realignment
- *10:30 Coffee break*
- 11:00 Base recalibration (BQSR)
- *12:00 Lunch break*
- 13:00 Hands-on session

Approximate schedule – timing may vary depending on Q&A

Day 3: Germline variant discovery

- 9:00 Recap of Day 2 and Q&A
- 9:15 Introduction to joint variant discovery
- 9:45 Variant calling and joint genotyping
- *10:30 Coffee break*
- 11:00 Variant recalibration (VQSR)
- 11:45 Manual filtration
- *12:00 Lunch break*
- 13:00 Hands-on session

Approximate schedule – timing may vary depending on Q&A

Day 4: Somatic variant discovery

- 9:00 Recap of Day 3 and Q&A
- 9:15 Introduction to somatic variant discovery
- 10:00 Estimation of contamination with ContEst
- *10:15 Coffee break*
- 10:45 Variant calling with MuTect
- 11:30 Rescuing TiN variants and eliminating artifacts
- *12:00 Lunch break*
- 13:00 Hands-on session

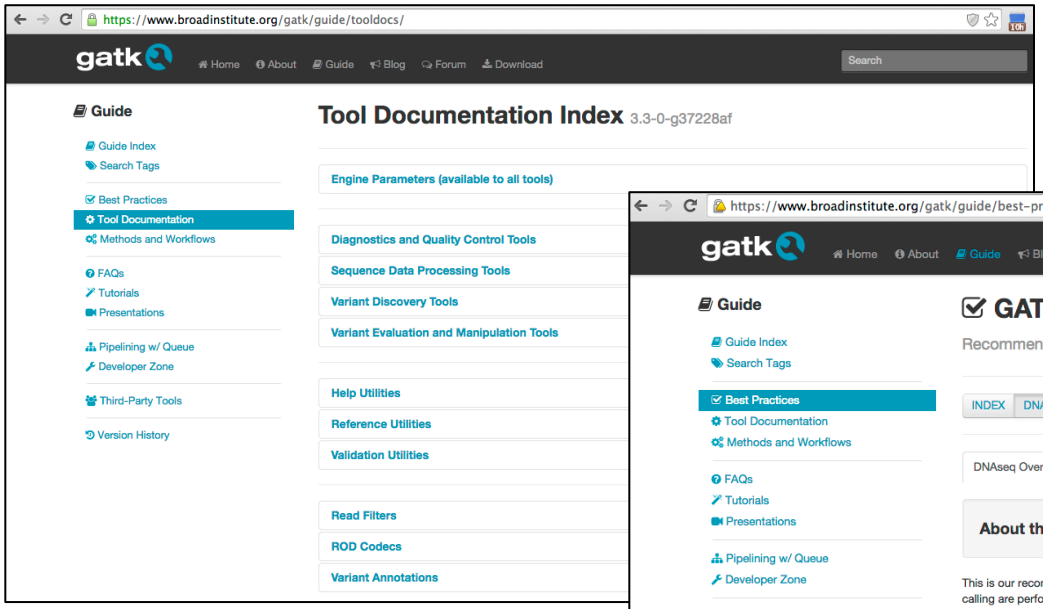
Approximate schedule – timing may vary depending on Q&A

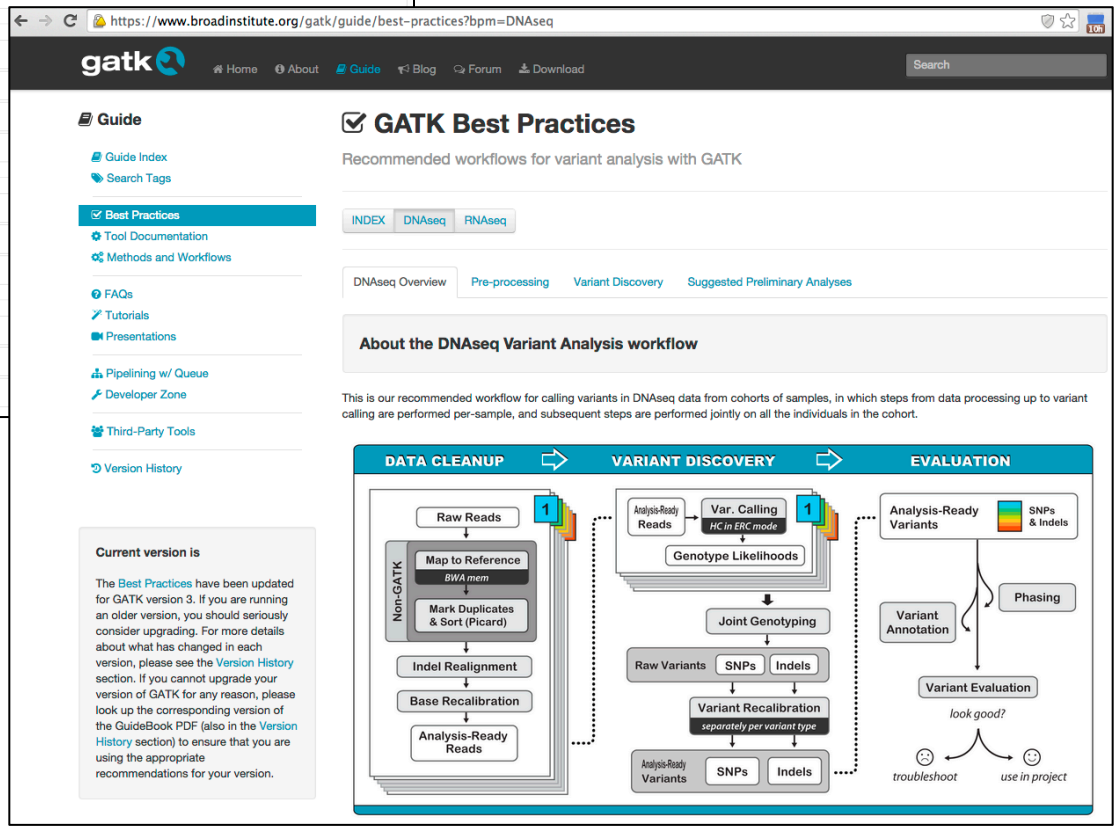
Day 5: Somatic variant discovery

- 9:00 Recap of Day 3 and Q&A
- 9:15 Introduction to working with variants
- 9:45 Genotype refinement
- *10:15 Coffee break*
- 10:45 Evaluating variants
- 11:30 Annotating variants
- *12:00 Lunch break*
- 13:00 Hands-on session

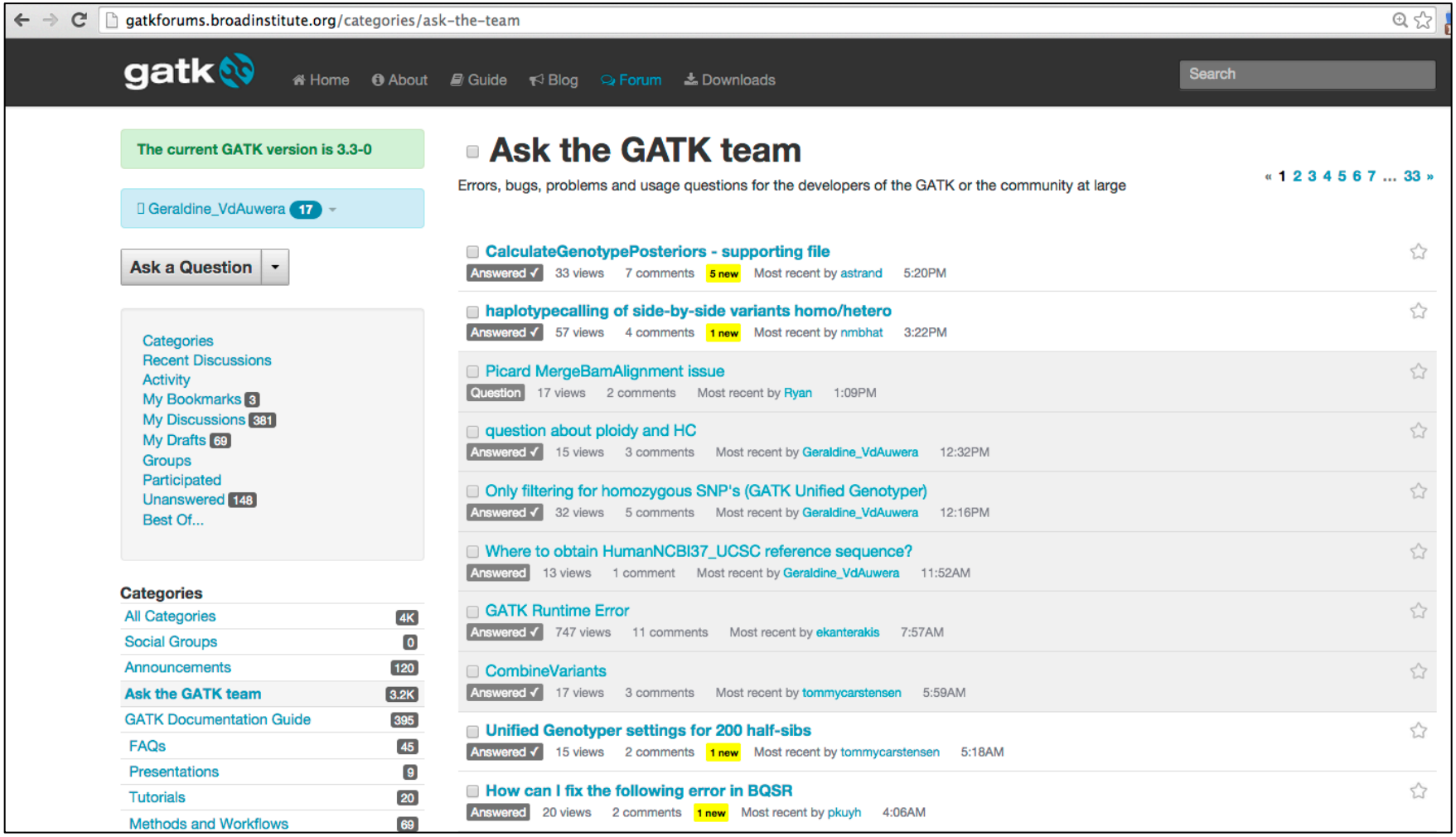
Approximate schedule – timing may vary depending on Q&A

Documentation is at https://www.broadinstitute.org/gatk/guide/





See the support forum for questions and bug reports



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