http://iseqtools.org/

GATK Best Practices for Variant Discovery

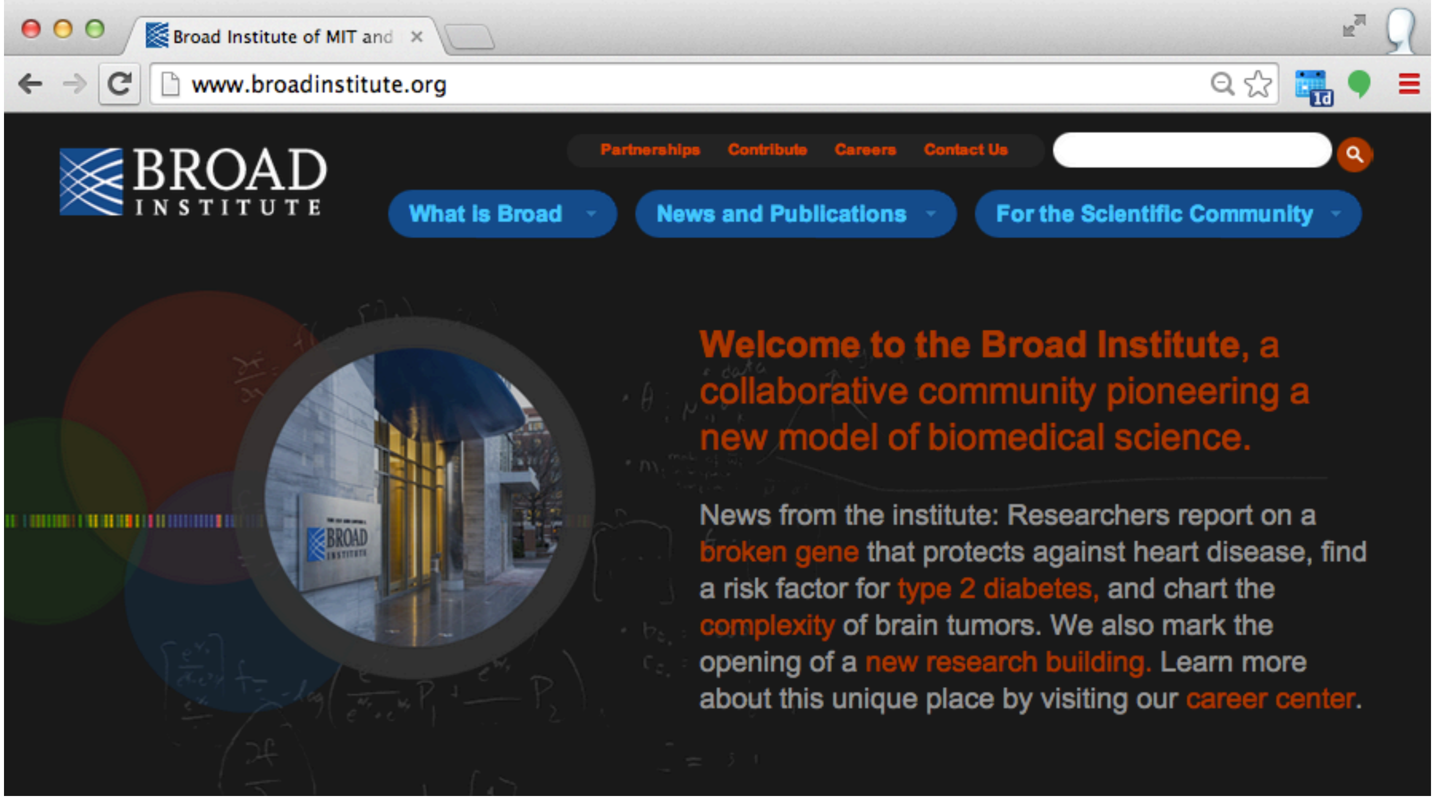
UCLA, Los Angeles CA, USA
2-4 Mar, 2016

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

@gatk_dev

What / who is the Broad Institute?

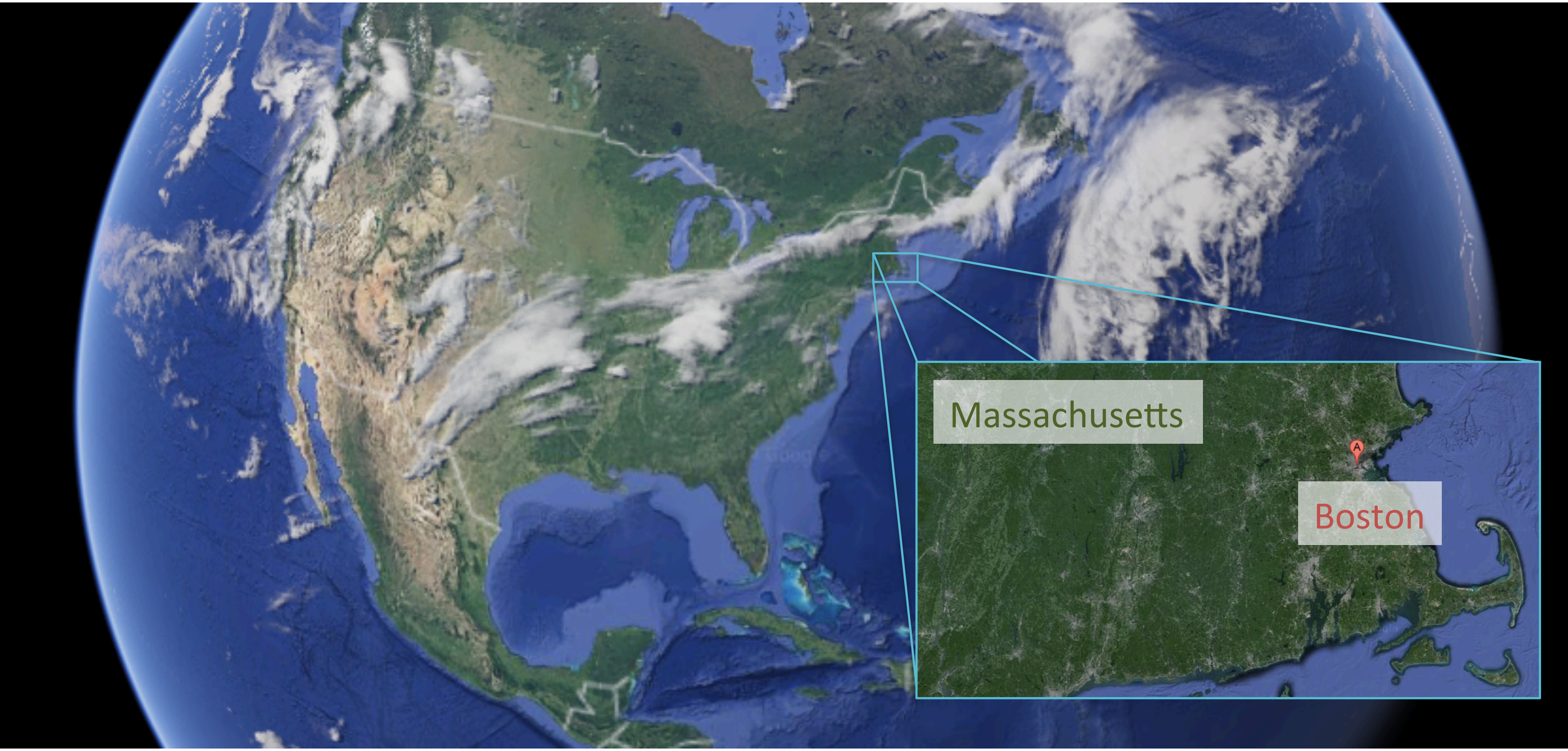
- Spinoff of Harvard & MIT -- Eric Lander and philanthropists Eli & Edyth Broad
- Use the full power of genomics to transform the understanding and treatment of disease



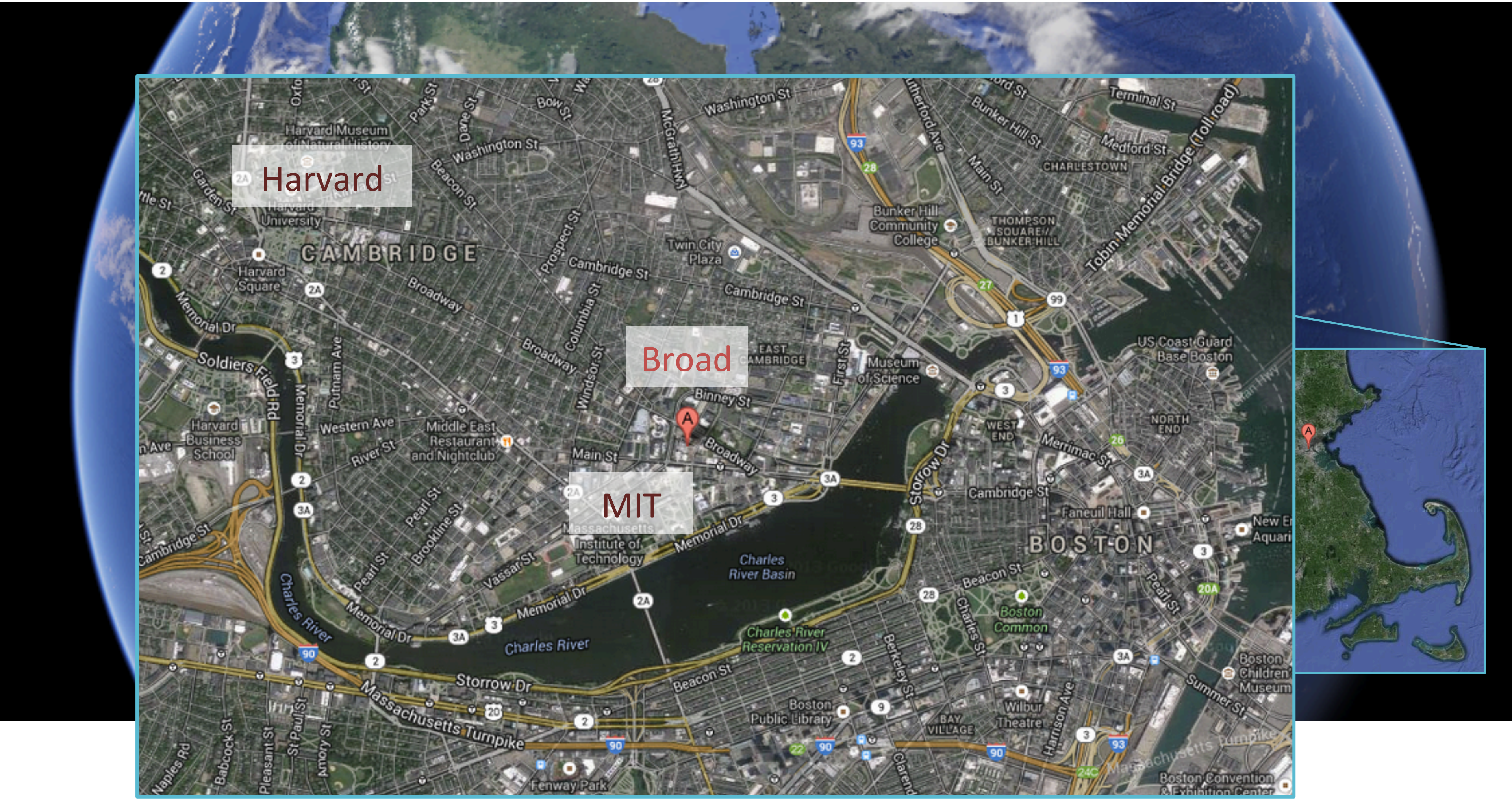
Welcome to the Broad Institute, a collaborative community pioneering a new model of biomedical science.

News from the institute: Researchers report on a broken gene that protects against heart disease, find a risk factor for type 2 diabetes, and chart the complexity of brain tumors. We also mark the opening of a new research building. Learn more about this unique place by visiting our career center.

Where in the world is the Broad?



Where in the world is the Broad?



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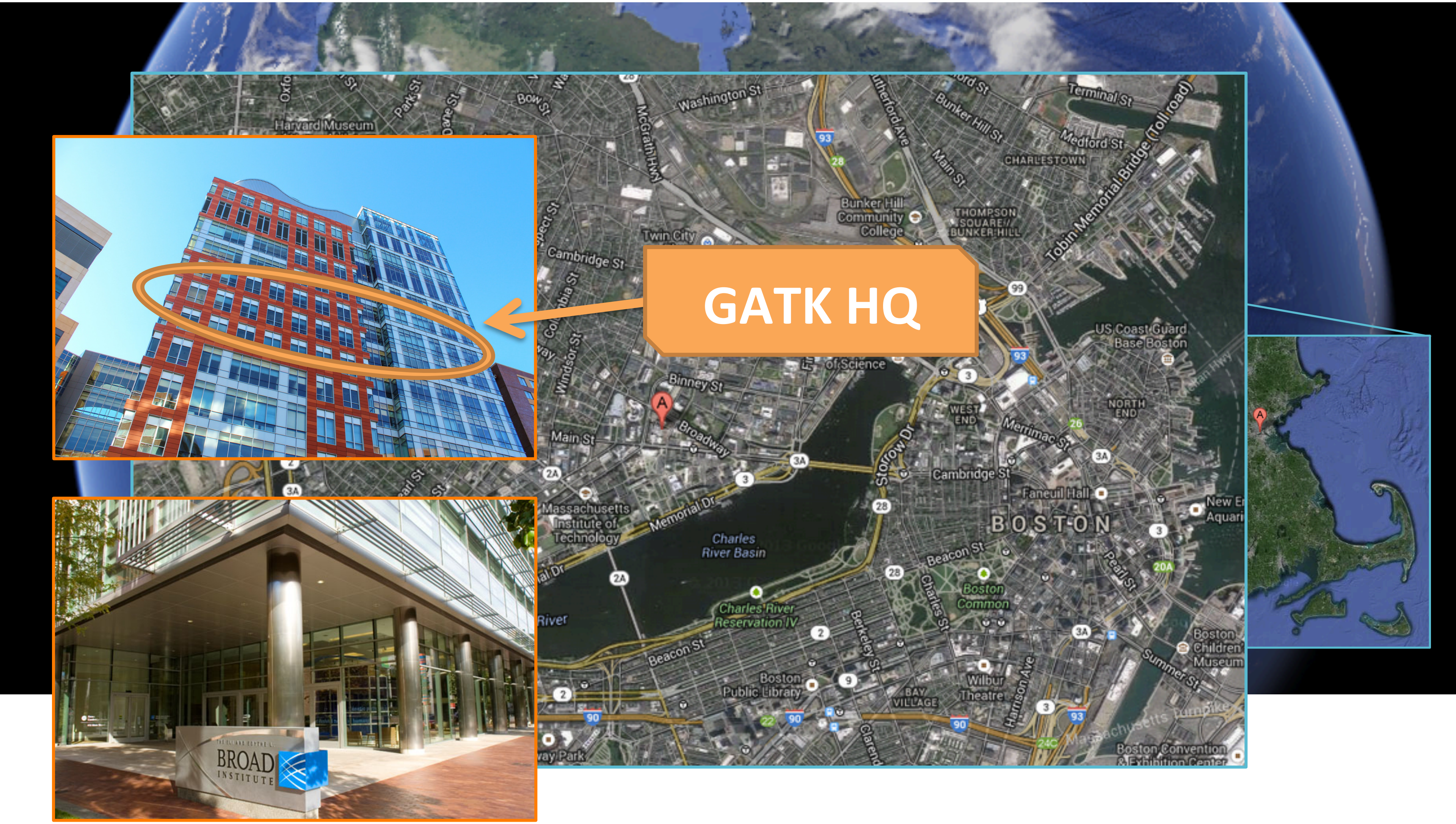


GATK HQ

Data Science & Data Engineering @ Broad

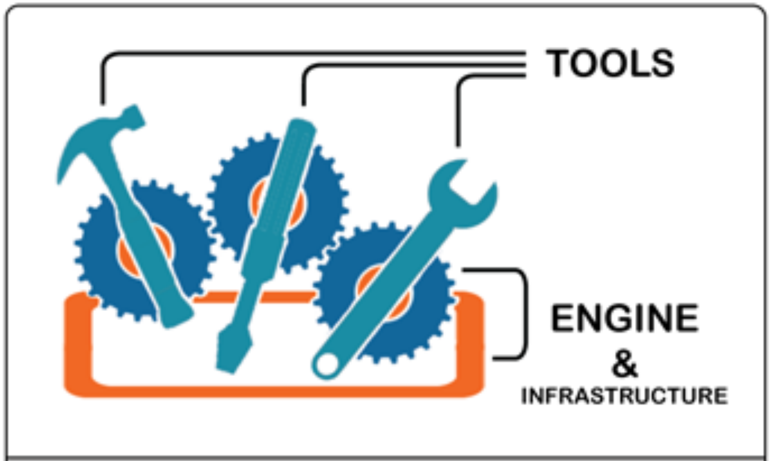
A new organization bringing together software engineers, computational biologists, and computing infrastructure specialists.

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

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

GATK = Genome Analysis Toolkit

- **Toolkit** focused on variant discovery (SNP & indel)
- **Components:**
  - Engine and infrastructure
  - Tools (walkers)

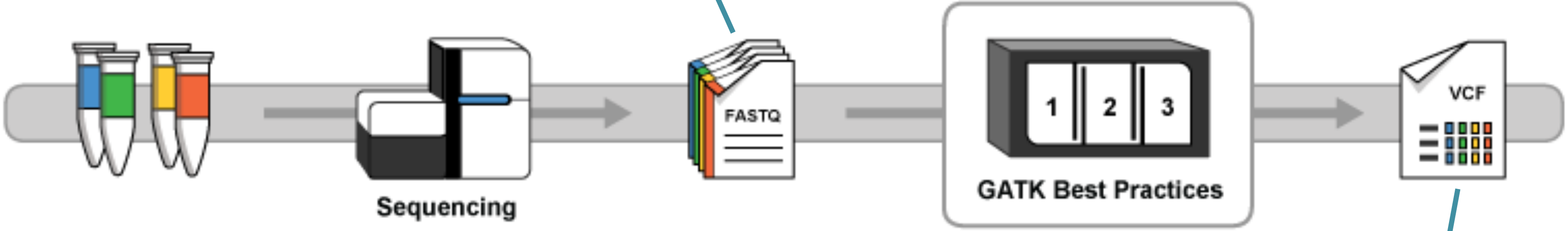


The core GATK components

- Also a **programming framework** for developing genome analysis software

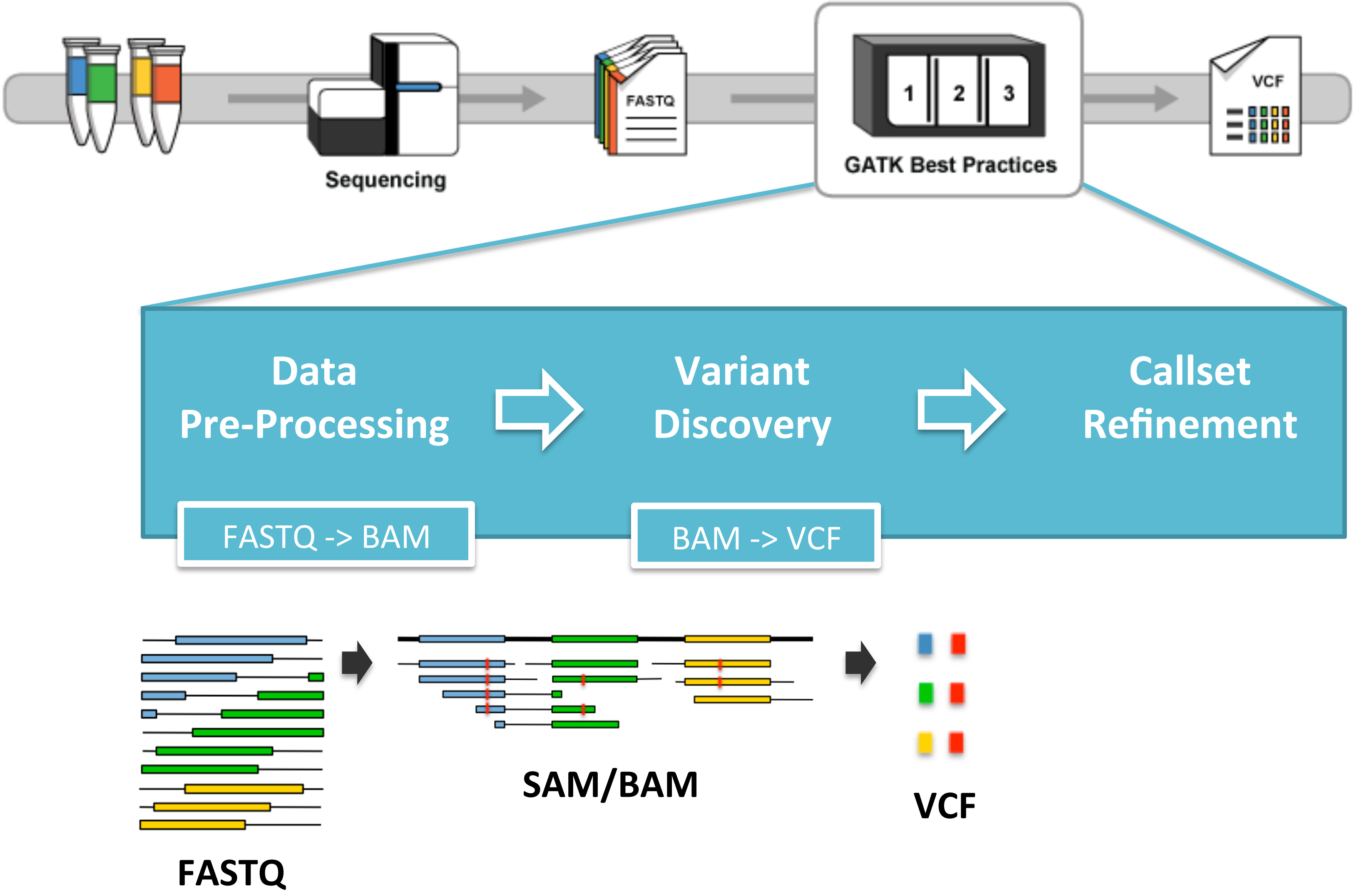
Variant discovery = identify **variants** in sequencing data

RAW READS
SEQUENCE DATA



VARIANTS

GATK Best Practices = complete **reads-to-variants** workflows

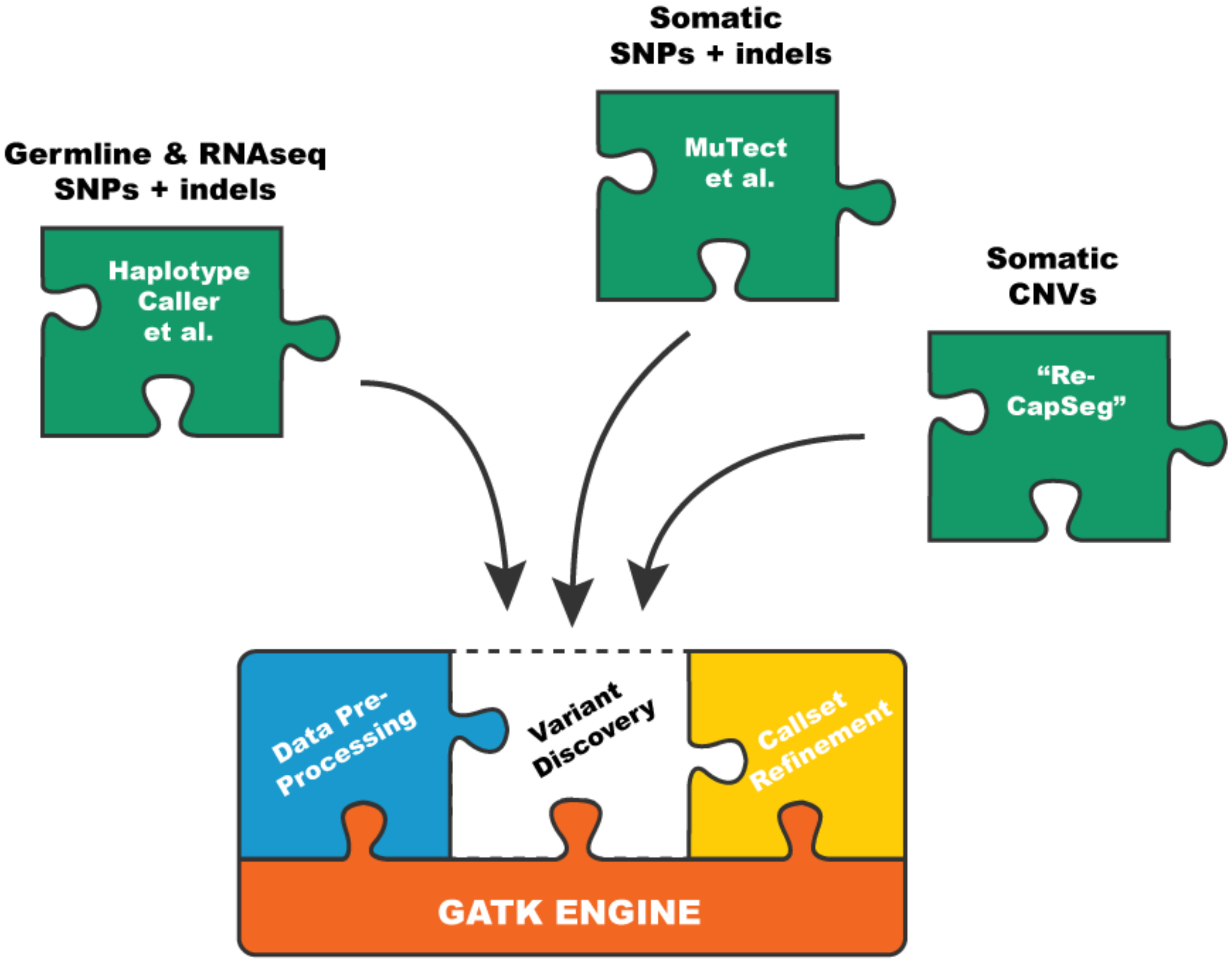


Data Pre-Processing → Variant Discovery → Callset Refinement

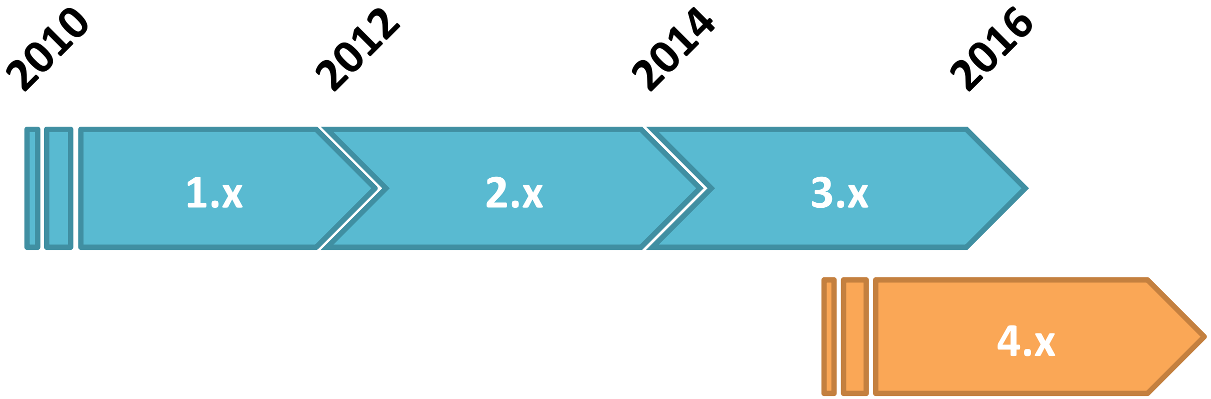
FASTQ -> BAM

BAM -> VCF

Expanding ecosystem of modular Best Practices workflows



GATK development roadmap



Alpha GATK 4 : cloud-friendly and more scalable (Apache Spark)
+ extended functionality (CNVs, Picard)

https://github.com/broadinstitute/gatk

Workshop agenda

Day 1

9 am – 10:30 am
Introduction to Variant Discovery

10:30 am – noon
Pre-processing methods

Day 2

9 am – noon
Germline variant discovery methods

Day 3

9 am – noon
Somatic variant discovery methods

Afternoons

2 pm – 5 pm
Hands-on tutorials