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Best Practices for variant calling with the GATK

Cambridge, MA, 4-5 Dec, 2012

Genome Sequencing and Analysis
Medical and Population Genetics
Broad Institute of Harvard and MIT
http://www.broadinstitute.org/gatk/

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General program

Day 1: Tuesday 4 Dec

Morning (9 AM – 1 PM)

Best Practices: From FastQ to analysis-ready BAM files

Afternoon (2 PM – 5 PM)

- Hands-on GATK
  (Board room)

OR

- Add-on analyses
  - 2 PM: GenomeSTRiP (B. Handsaker)
  - 3 PM: XHMM (M. Fromer)

Day 2: Wednesday 5 Dec

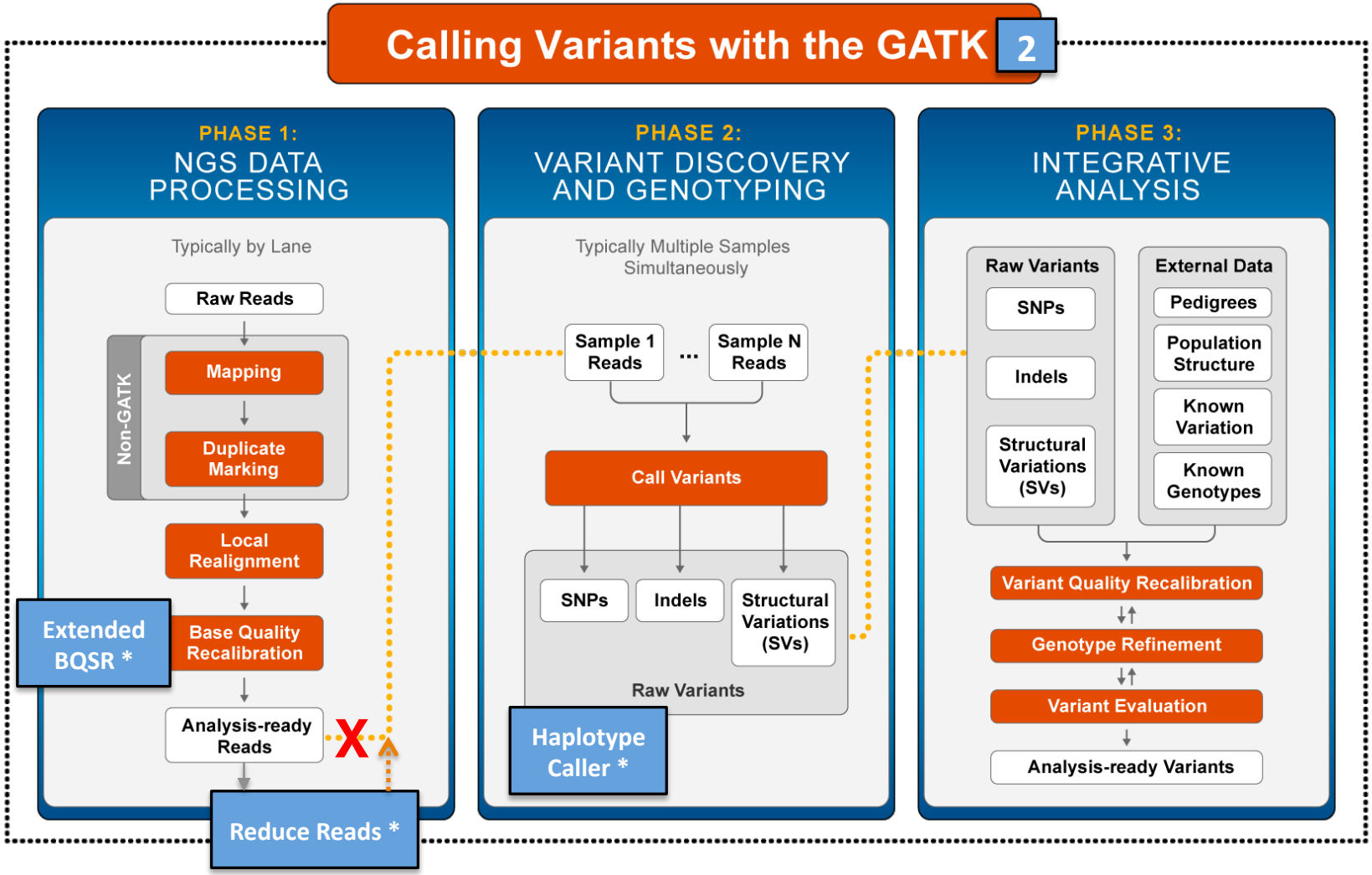
Morning (9 AM – 1 PM)

Best Practices: From analysis-ready BAM to analysis-ready VCF files

Afternoon (2 PM – 5 PM)

- Hands-on GATK
  (Nile room @ 301 Binney)

Topics covered in the main track



* New tools or functionalities not available in GATK-Lite

Topics covered in the hands-on session

- Building a command-line
- Running analysis tools
- Understanding console messages
- Dealing with errors
- Using engine arguments
- Finding relevant information in the documentation
- Using complementary third-party utilities

Please check in with us during coffee break!

Genome Sequencing and Analysis (GSA)

Mark De Pristo






Speakers (in order of appearance)

Géraldine Van der Auwera
Eric Banks
Ami Levy-Moonshine
Mauricio Carneiro
Ryan Poplin
Guillermo del Angel
Chris Hartl (not pictured)

Other GSA members

Khalid Shakir
David Roazen
Joel Thibault

Best Practices:
From analysis-ready BAM files to analysis-ready VCF files

• 9:00 AM	Opening address	Geraldine
• 9:15 AM	Variant calling (UG & HC)	Ryan & Guillermo
• 10:15 AM	VQSR	Ryan
• *11:00 AM*	*Coffee break*	
• 11:30 AM	Genotype refinement & Functional Annotation	Eric
• 12:00 PM	Variant manipulation & analysis	Chris
• 12:45 PM	Closing comments and exit poll	Geraldine
• *1:00 PM*	*Lunch break*	