

# **Overview of the Genotyping-By-Sequencing Technology and Its Applications**

Qi Sun

Computational Biology Service Unit

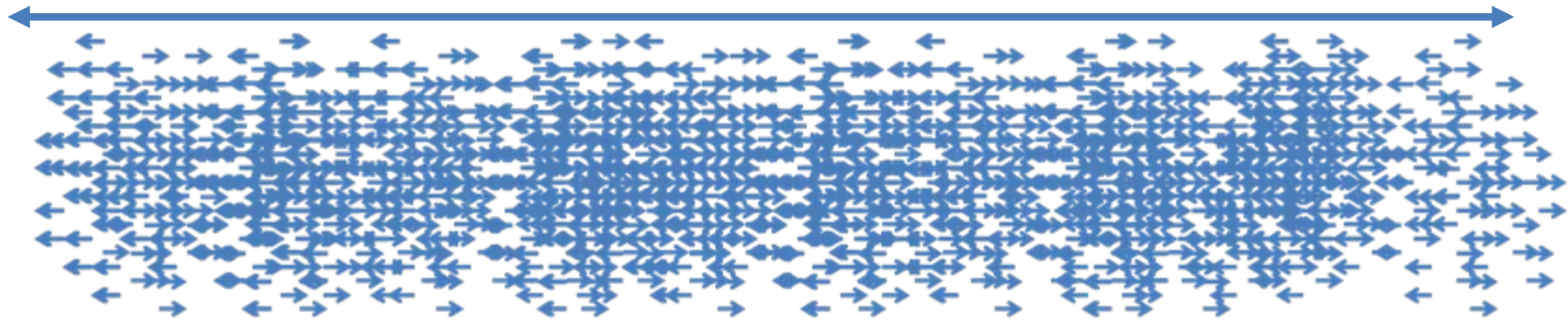
Cornell University

# Whole Genome Shotgun (WGS)

*De novo* Assembly

Coverage: 100x, (multiple libraries with different insertion size)

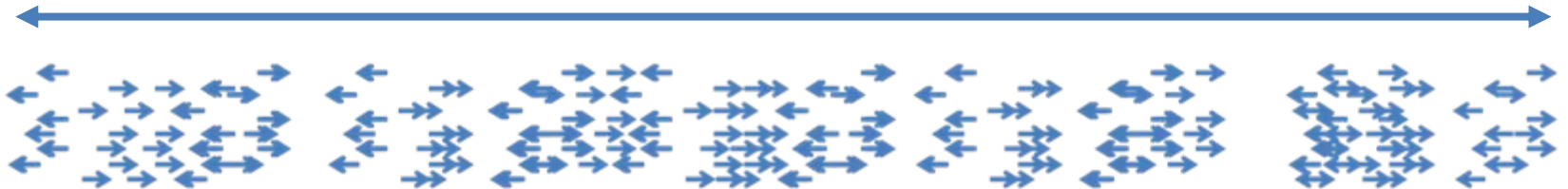
Cost: ~\$25,000



SNP/INDEL detection through alignment to reference genome

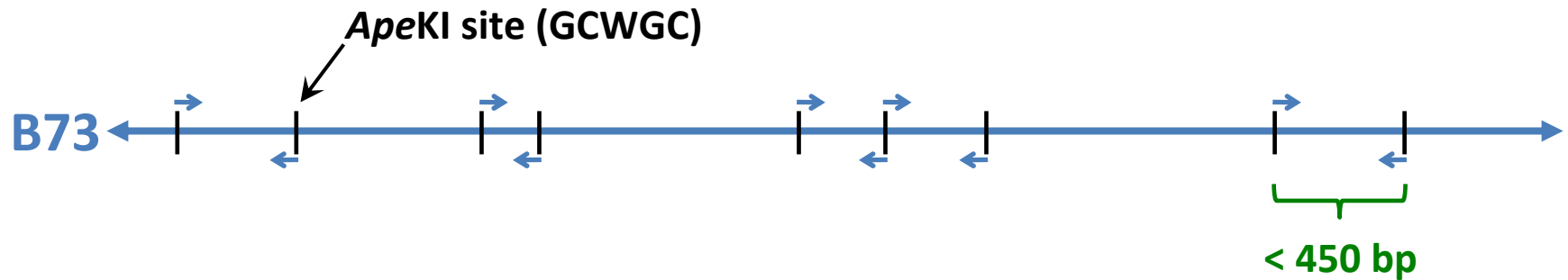
Coverage: 1 to 10x

Cost: \$250 to \$25,00



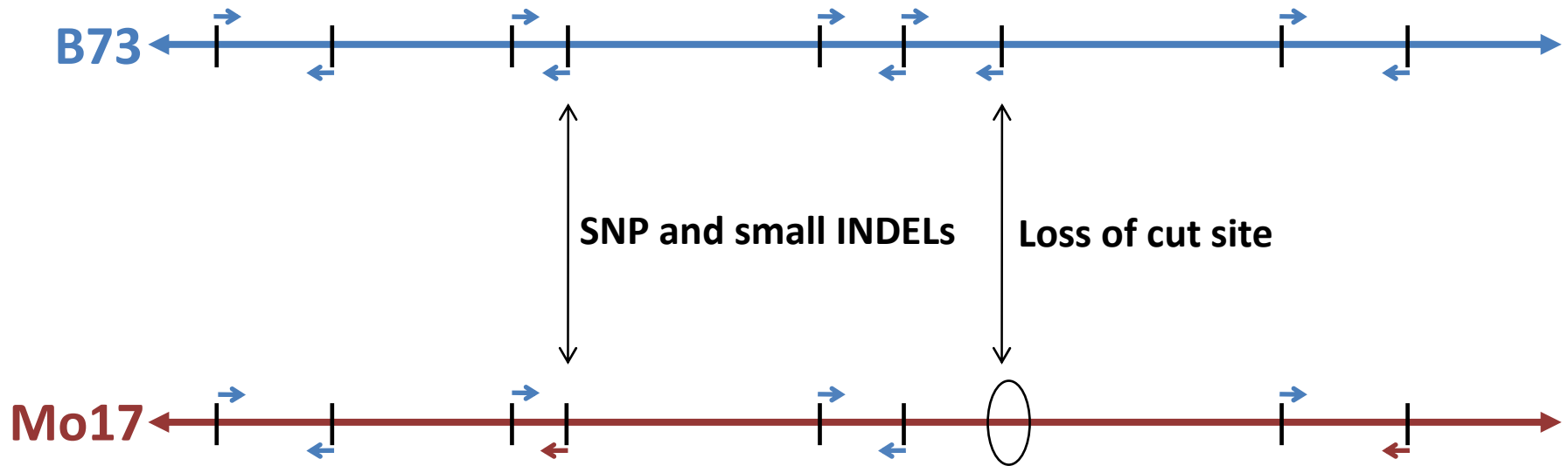
# Genotyping by Sequencing (GBS)

(→) 64-base sequence tag

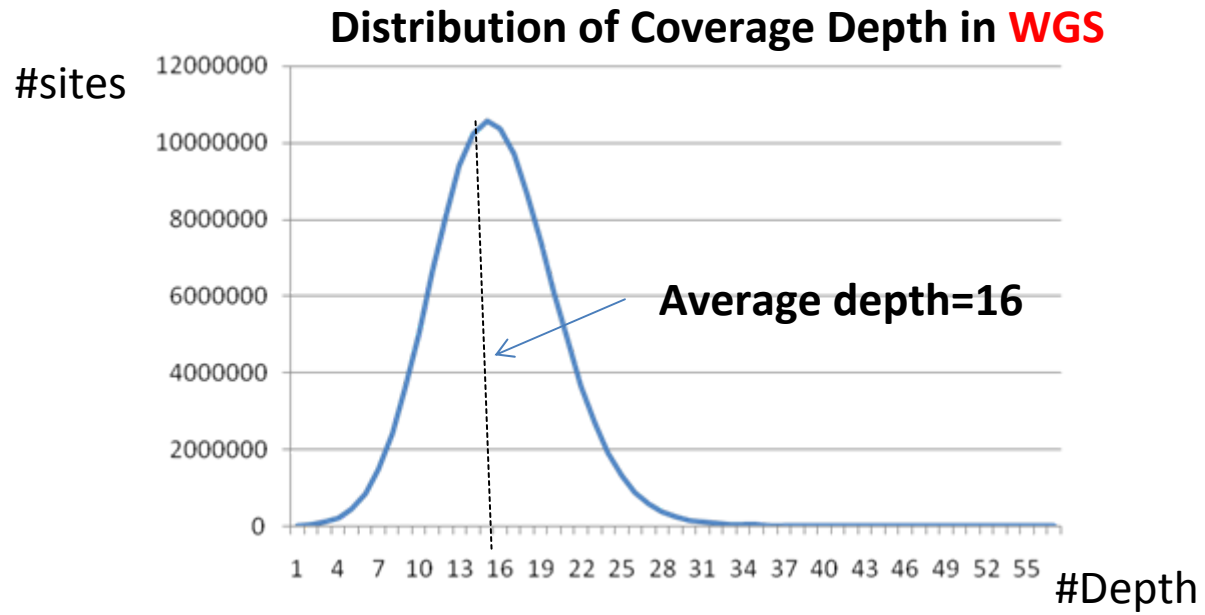
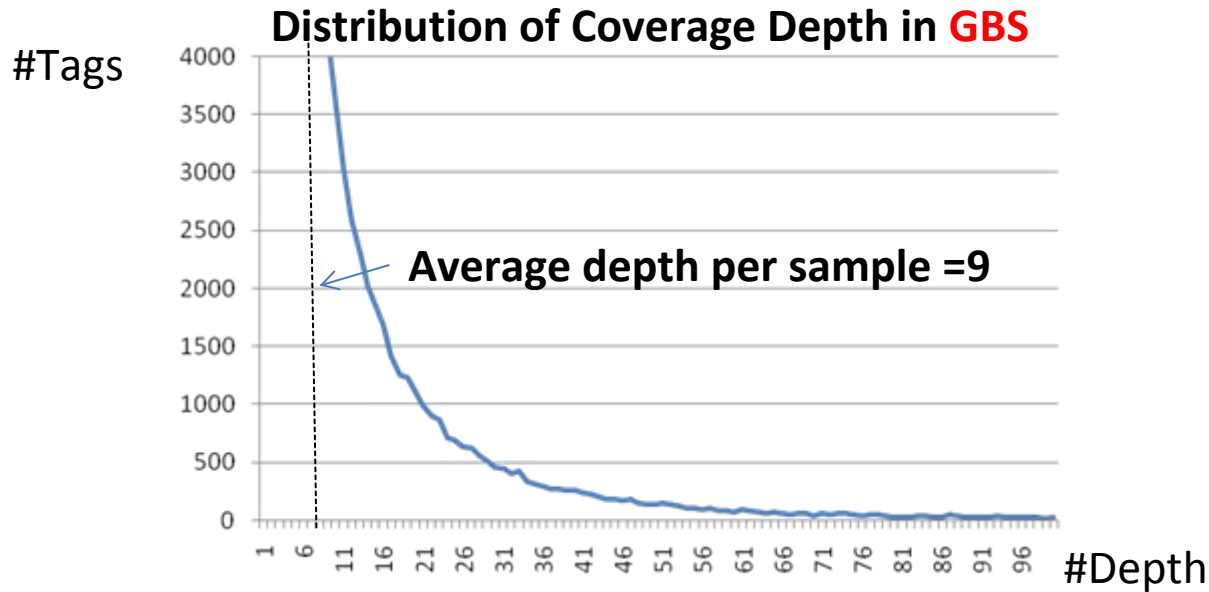


- **Reduced genome representation;**
- **Reads can be aligned without reference genome;**
- **Low coverage per individual, coverage achieved by combining the reads from a population.**

# Identification of markers with/without the reference genome



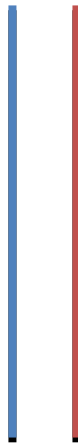
# Difference of Coverage depth between GBS and Whole Genome Sequencing data



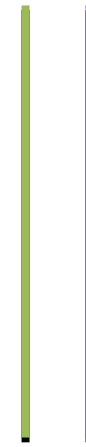
## GBS on Grape F1 population

chardonnay

rupestrisB38



X



**86 F1 population**

# **GBS of the F1 population**

**160 million reads**

**0.7 million tags**

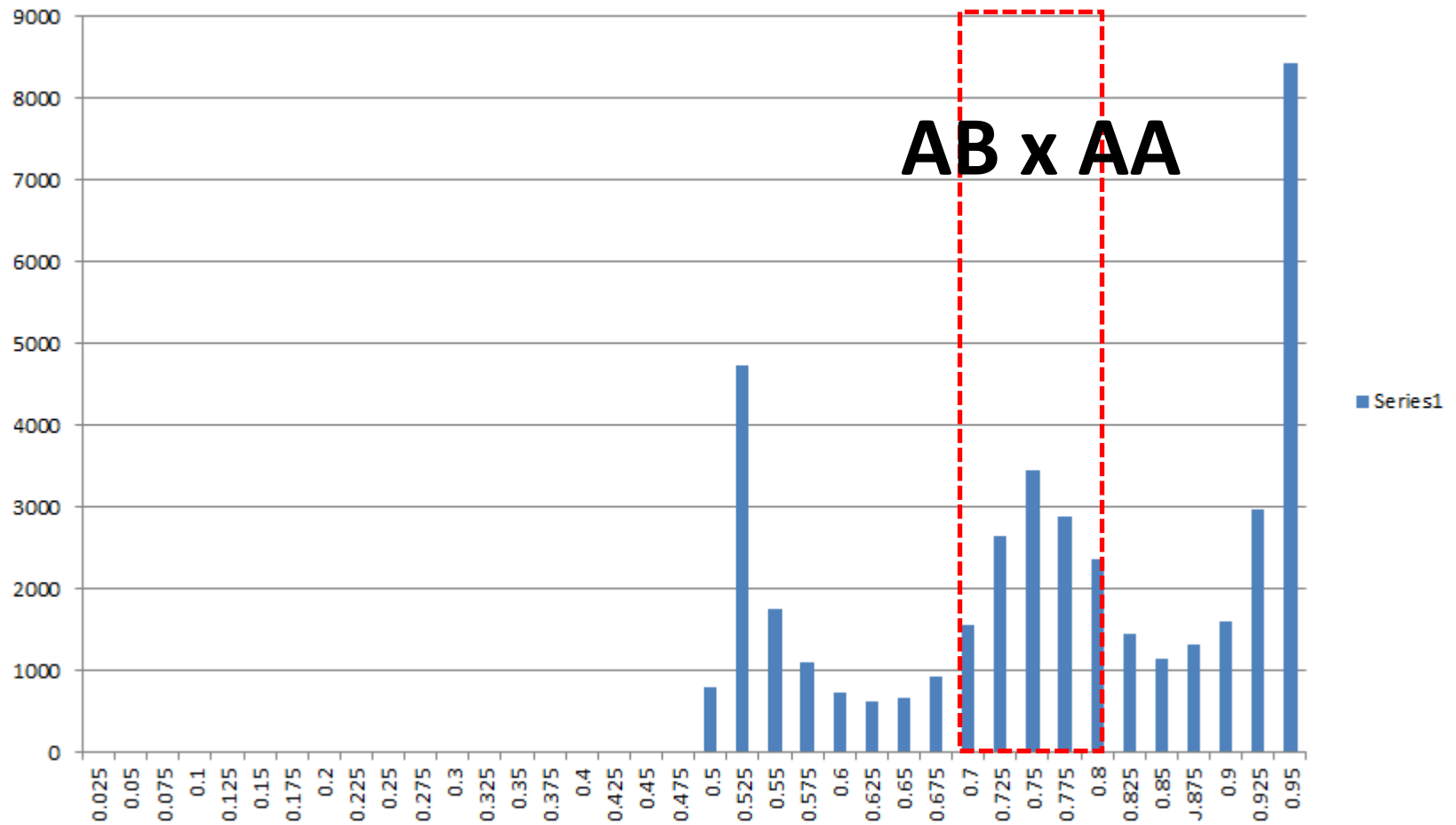
**10k biallelic markers**

**i.80% coverage**

**ii.MAF 25%**

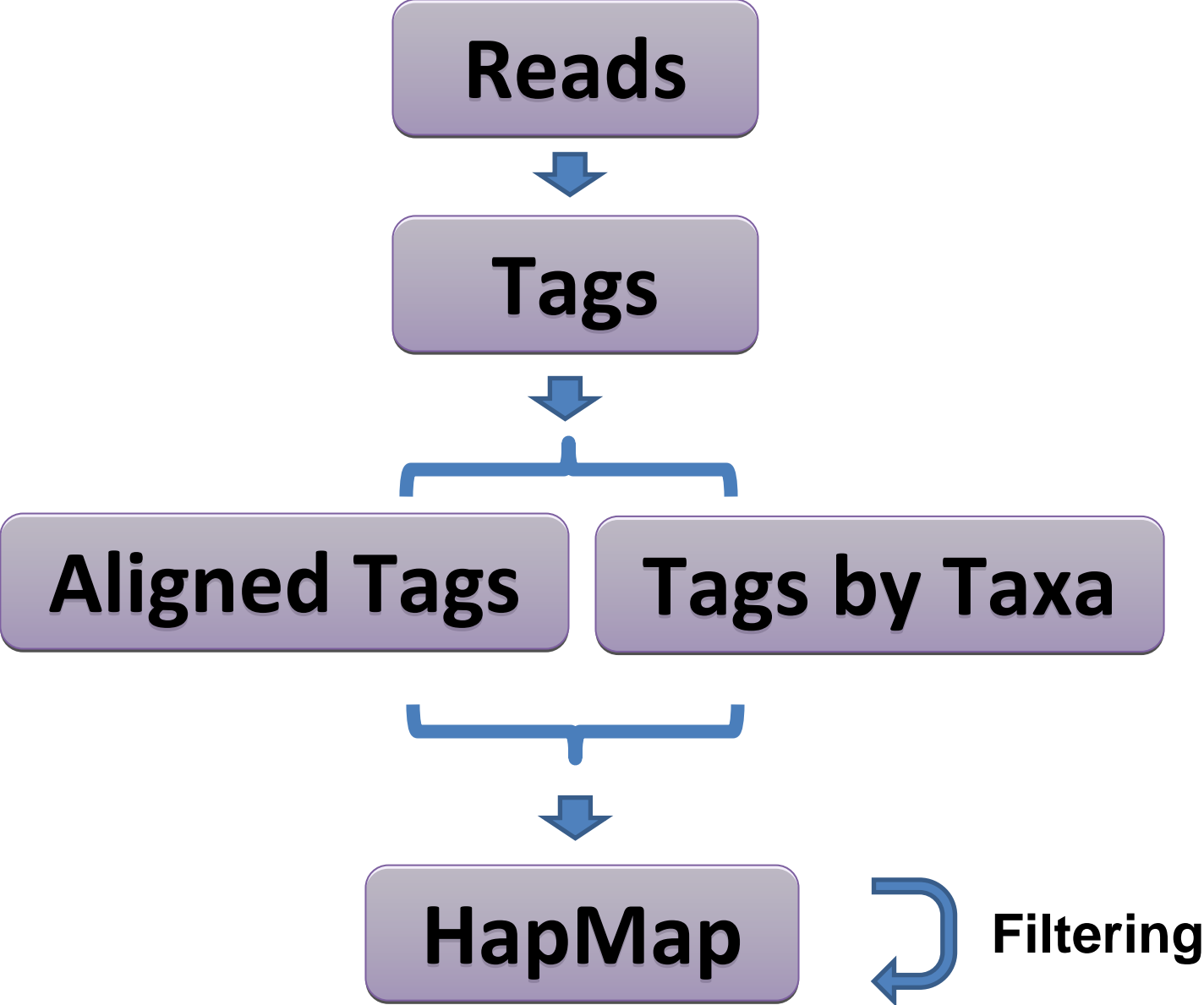
**iii.physical map match genetic map)**

# Strategy for Map Construction

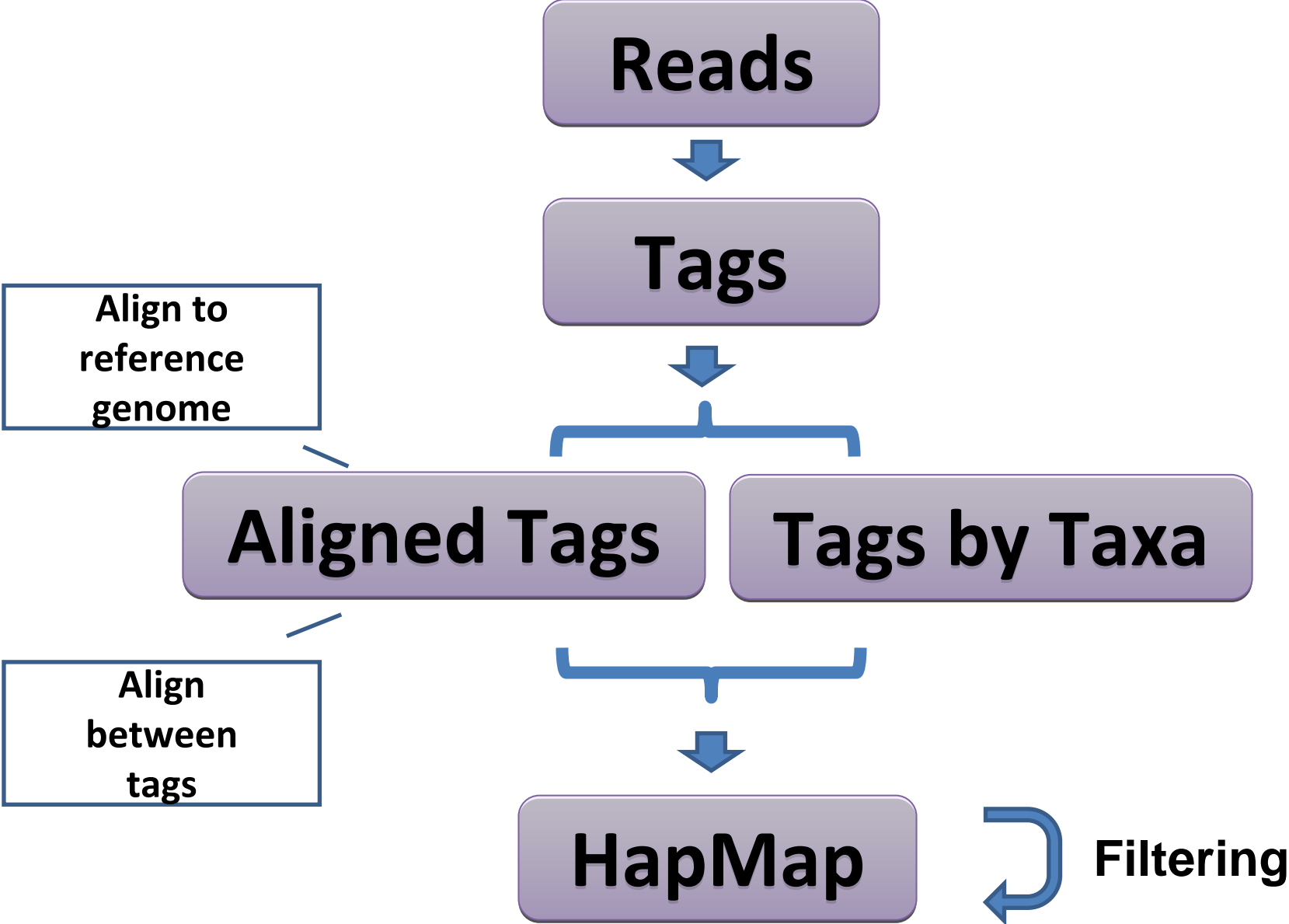




# GBS Data Analysis Pipeline



# GBS Data Analysis Pipeline



# Applications of the GBS markers

## Goal of year 1:

**1.Marker discovery**

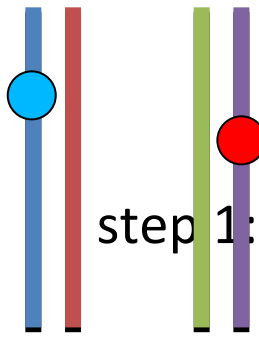
**2.Linkage map construction.**

**3.QTL mapping.**

## Goal of year 2 -3:

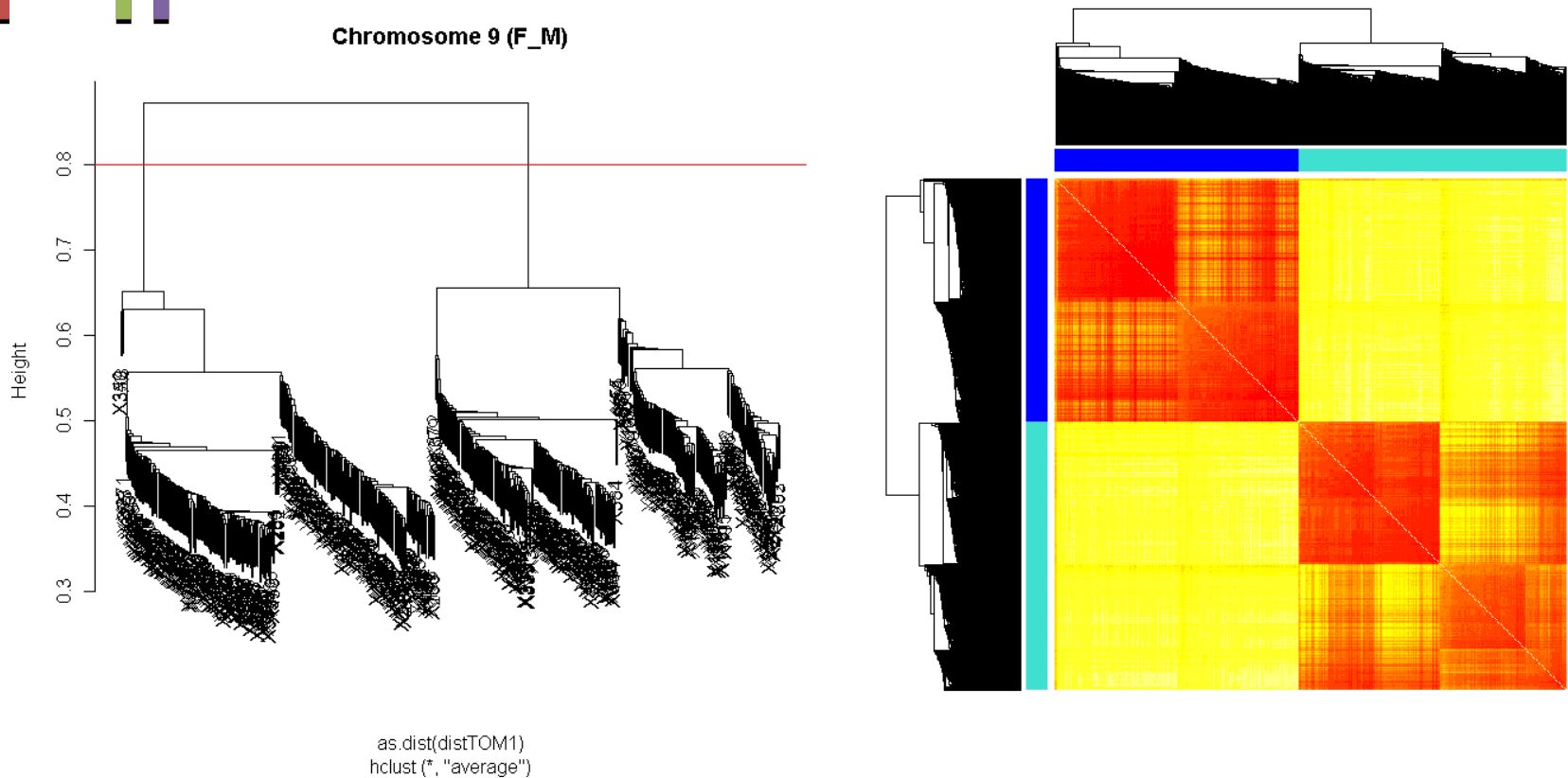
**1.HapMap construction. Integration of GBS maps with genome sequences.**

**2.Genomic selection.**

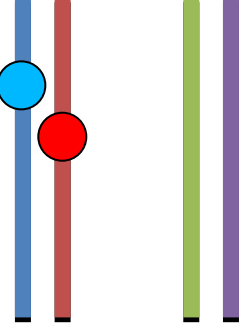


# Strategy for Map Construction

step 1: For each marker, assign the minor alleles to one of the parents



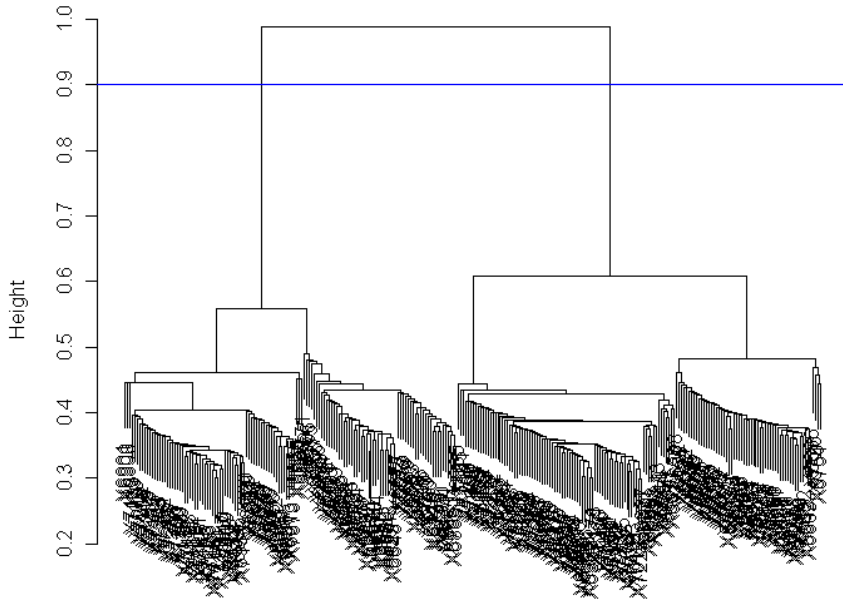
Chromosomal assignments of 10% to 50% of the markers are inconsistent with the released genome assembly (Pinot Noir)



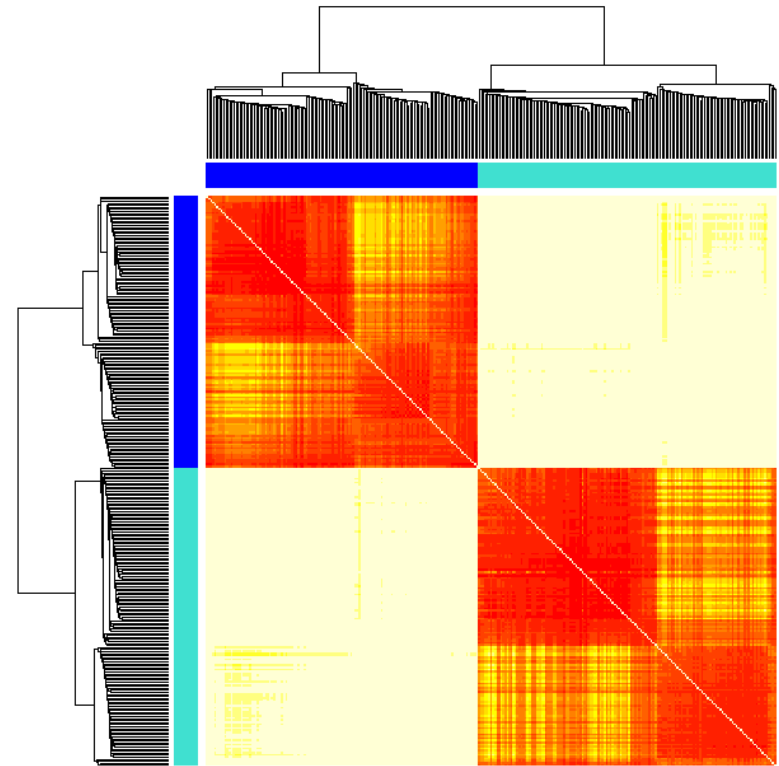
# Strategy for Map Construction

step 2: phasing of the heterozygous markers

chromosome 9 (F)



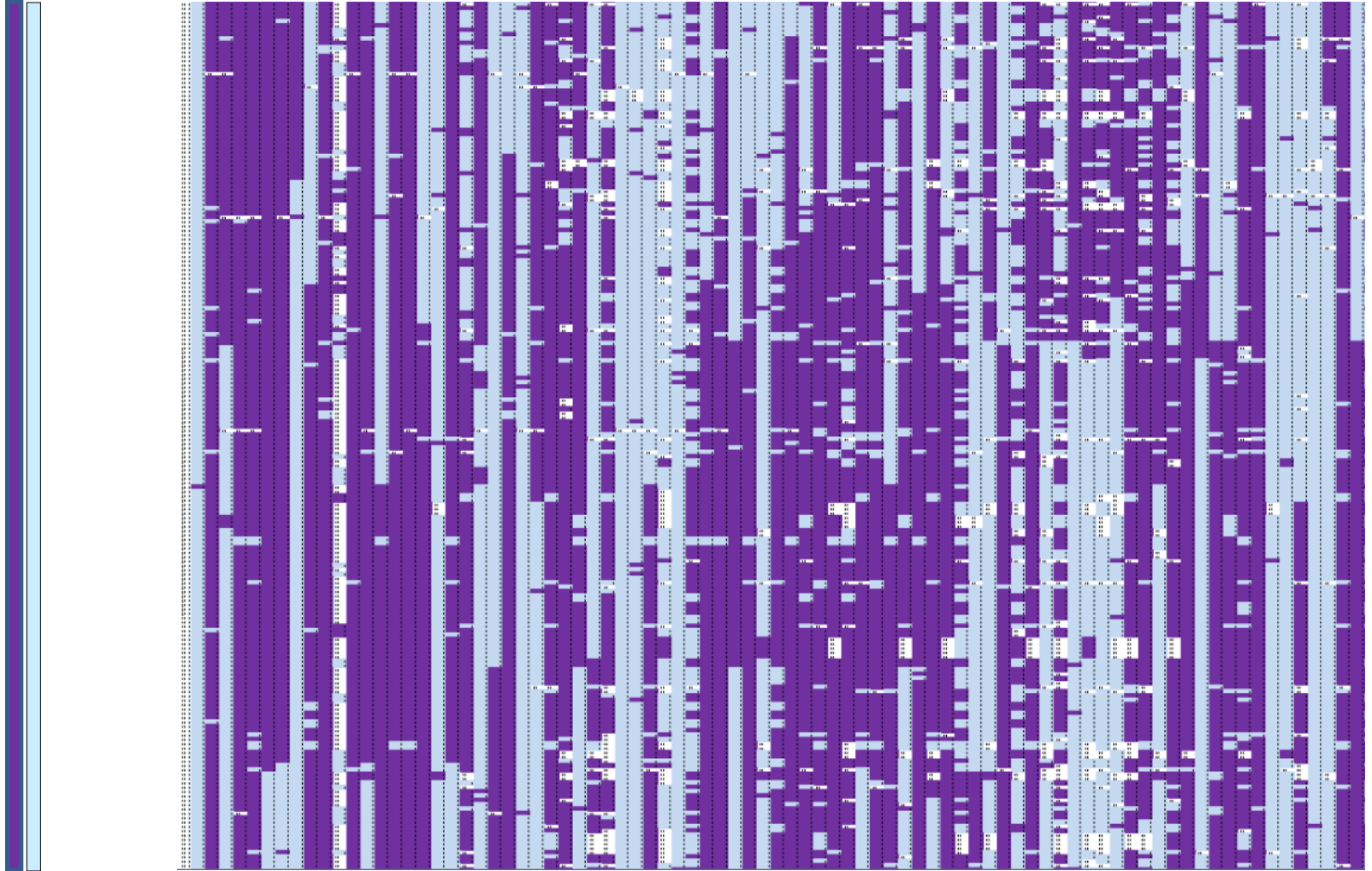
```
as.dist(distTOM1)  
hclust("average")
```



### Step 3. Error correction, ordering and imputation (work in progress)

Parent 1

86 F1 progenies



Map before imputation


## 10 K biallelic markers can be genetically mapped

12386	Markers with 20% or less missing data
8259	Genetically and physically mapped to the same chromosome
2495	Genetically and physically mapped to different chromosomes
1632	not mapable

# GBS Workshop

## Ithaca, NY Feb.16-17, 2012

Buckler lab - IGD - CBSU



Cornell University  
Life Sciences Core Laboratories Center  
Computational Biology Service Unit

SEARCH CORNELL:

Pages  People [more options](#)

[HOME](#) [Mission and Services](#) [BioHPC](#) [Staff](#) [Publications and Outreach](#) [CBSU/3CPG BioHPC Lab](#) [Contact us](#) [Forum](#) [Mail list](#)

### Genotyping-by-Sequencing (GBS)

February 16-17 2012

[Click here to register for this workshop](#)

Cornell's Institute for Genomic Diversity and Computational Biology Service Unit are jointly offering a two-day workshop on genotyping-by-sequencing (GBS), a new method for generating large numbers of single nucleotide polymorphisms (SNPs) for use in genetic analyses. The workshop will be held at 655 Rhodes Hall, Cornell University, Ithaca, New York.


The first day of the workshop will comprise a series of lectures with emphasis on data analysis with current pipelines. On the second day, a computer lab will be available for analyzing raw GBS sequence data from several species. Participation in the computer lab is limited to 40 individuals.

#### Workshop Outline

[Session 1](#) 2/16/2012 9:30:00 AM 655 Rhodes Hall  
Lectures

[Session 2](#) 2/17/2012 9:30:00 AM 655 Rhodes Hall  
Computer Lab

For people coming to the workshop from outside Cornell, we have reserved a block of hotel rooms at the Best Western near campus. Call 607-272-6100 and request the GBS workshop rate (\$99) to reserve a room. Email [Theresa.tf12@cornell.edu](mailto:Theresa.tf12@cornell.edu) if problems or other logistical questions.

 [login](#) [BioHPC @ CBSU](#)

Google: GBS workshop



# Acknowledgement

## GBS Platform

Ed Buckler  
Jeff Glaubitz  
James Harriman  
Robert Elshire  
Sharon Mitchell

## VitisGen

Bruce Reisch  
Lance Cadle-Davidson  
Paola Barba

## Computational Biology Service Unit

Jarek Pillardy  
Robert Bukowski  
Minghui Wang

