

GVA 2020 Review

Attempts to add perspectives and additional resources.

Reminder of goals and consider how well they were met

- Teaching goals:
 - Teach the fundamentals of NGS variant analysis.
 - The wiki page
 - Provide context and exposure multiple types of data.
 - SE, PE, MP sequencing
 - Virus, bacteria, plasmid, human in different tutorials
 - Use example commands to familiarize you with variety of programs.
 - The wiki page
 - Provide resources to enable you to do analysis you haven't thought of yet.
 - The wiki page

Stages of NGS analysis

1

Biological
Question

2

Design &
Conduct
Experiment

3

Prepare NGS
Library &
Sequence

4

Sequencing
Analysis

Typical Stages of Variant Analysis

1

Read Quality
Control

2

Map Reads

3

Identify Variants

4

Visualize Variants

#1 most common question I get asked

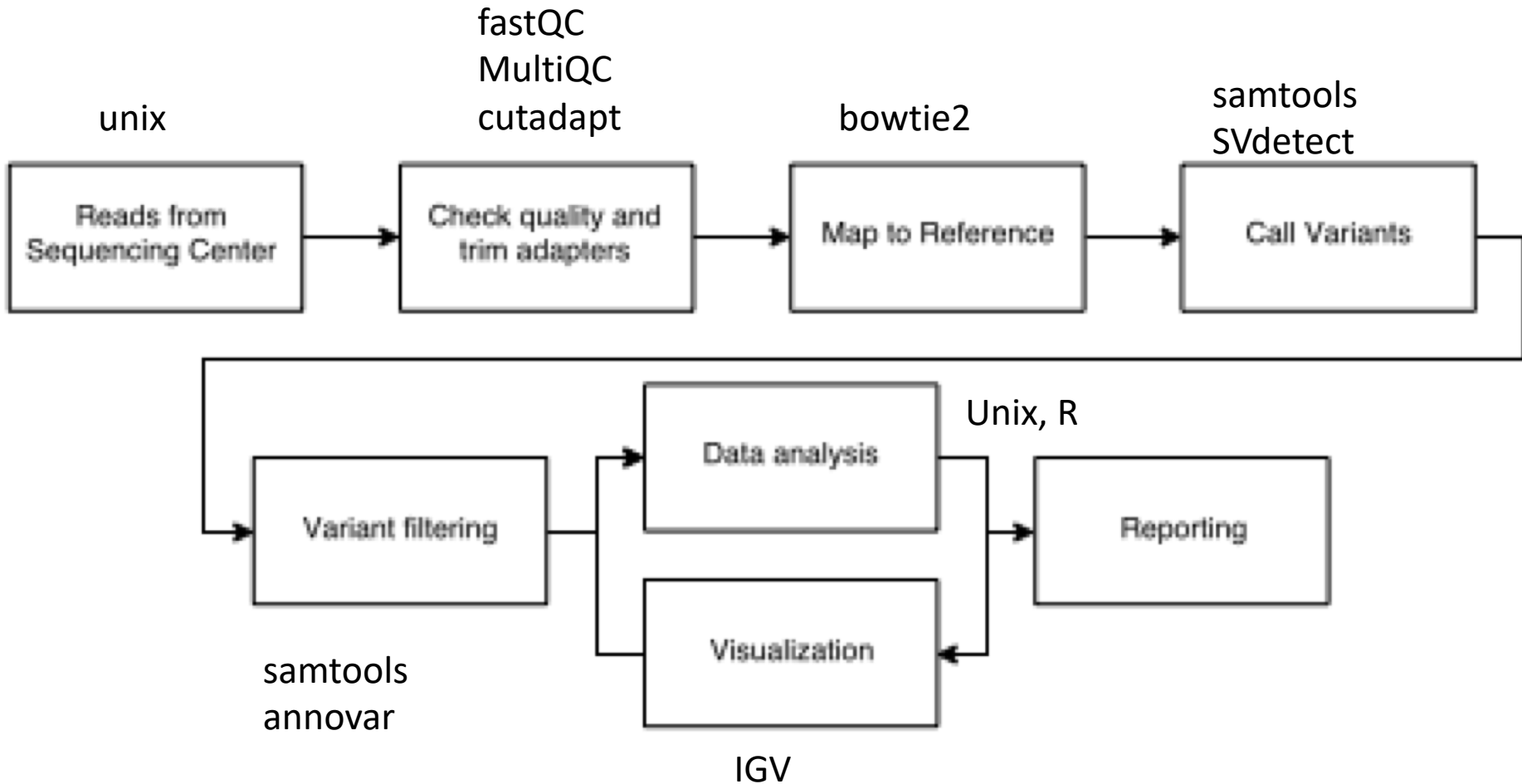
- How much sequencing do I need to do?
 - Most applications 30-50 fold coverage, higher for bacteria/small organisms because they smaller and cheaper.
- How do I change reads or lanes into coverage?

$$\text{Coverage} = \frac{(\text{Read Length}) \times (\text{Sequencing Type}) \times (\text{Number of Reads})}{\text{Size of Genome}}$$

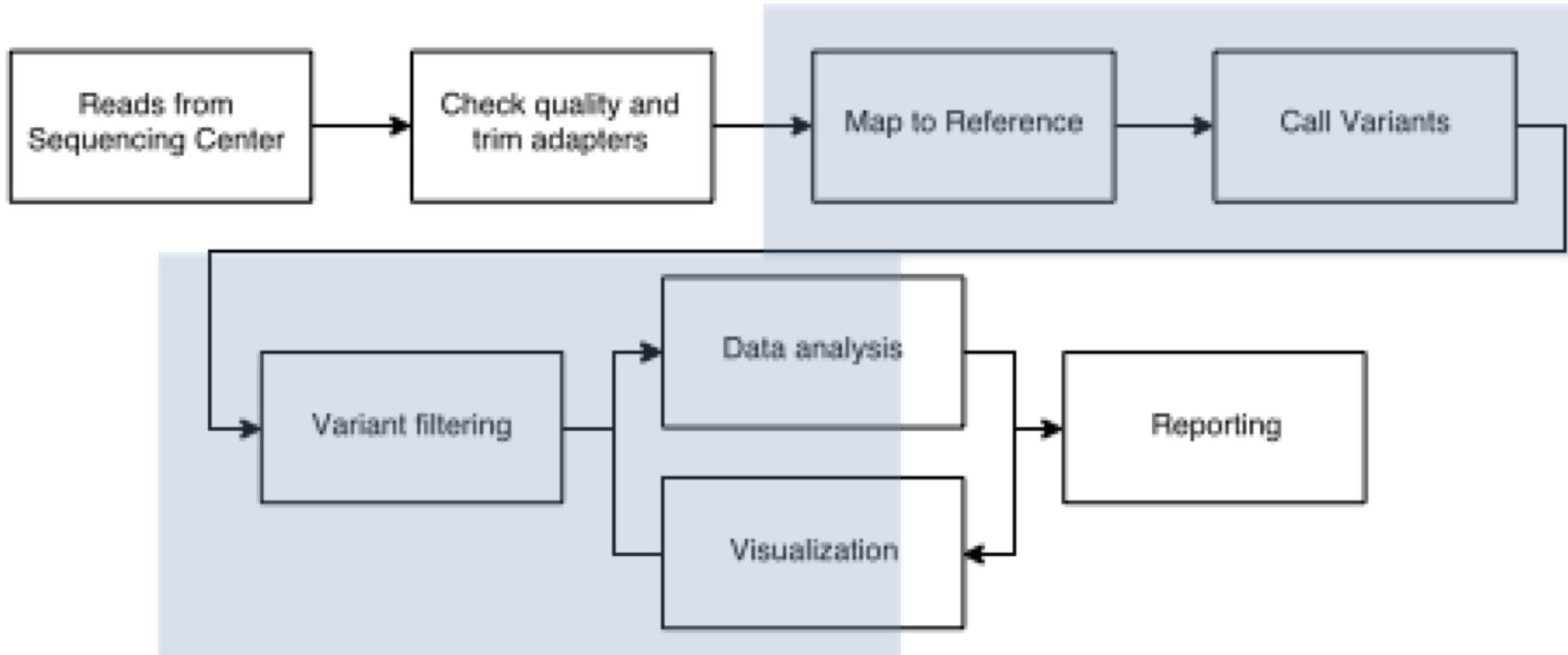
$$30 = \frac{150 \times 2 \text{ (if Pair end or 1 if single)} \times (\text{Number of Reads})}{\text{Size of Genome}}$$

- Min number of reads = ~10% of the genome length
 - If PE 150bp run.
- Max number of reads = ~30% of the genome length

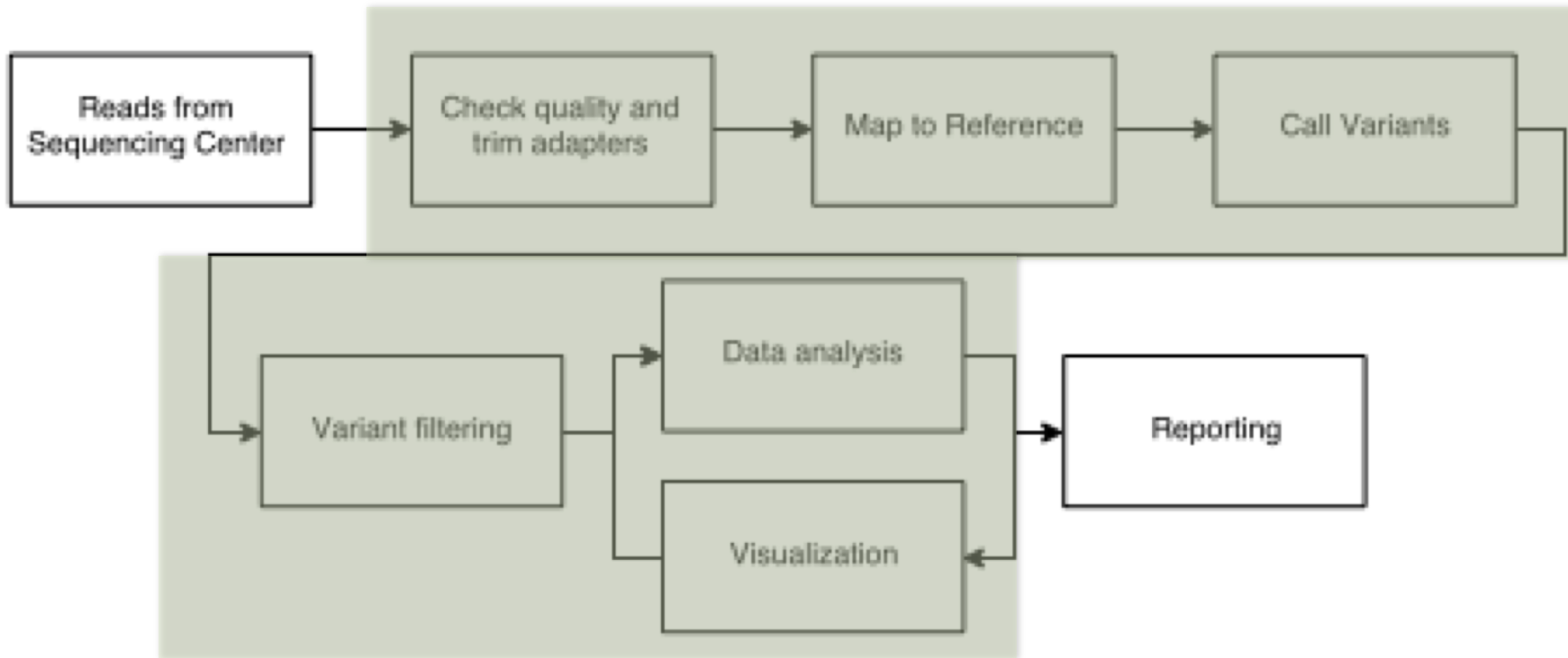
Steps for GVA



microbial all-in-one: breseq



eukaryotic all-in-one: GATK



Further Resources (online)

- Course wiki:
<https://wikis.utexas.edu/display/bioiteam/Genome+Variant+Analysis+Course+2020>
- Galaxy : <https://usegalaxy.org>
- Coursera: Genomic data science :
<https://www.coursera.org/specializations/genomic-data-science>
- edX: Python,R:
<https://www.edx.org/course/subject/computer-science>
- Course instructor. You have my email.

What's next

- Today, keep working on tutorials
 - Hint hint job submissions!
- Talk to me about what you don't understand about what we have done or why something was important or how it fits together.
- Keep eye out for email from me and from CCBB to review your experience, I really appreciate feedback, it's the only way to make this course better for other people.
- Soon, start analyzing your own data.

Additional tutorials available

- Novel DNA Identification had some additional information added to the discussion of results and next step sections
- Advanced mapping and breseq with mult. Refs
 - Tie in well with the Novel DNA tutorial.



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