31 Mar 2025

Sarek_Mic

Advanced Molecular Detection
Southeast Region Bioinformatics

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Updates

- Complete BRR Needs Assessment if you haven't yet – the more responses, the better!
- BRR/BTL Training - Delayed this year after changing from ABiL to UGA as our tutorial provider. Please update your ELC Milestones to notify them of this. We maintain contact with SaGE/OAMD team on these changes as well!

Overview

Purpose

- Analyze microorganisms such as Candida auris, SARS-CoV-2, TB, and more with references which result in SNP calling and annotation that enhances for genomic research.

Usage

- To support public health and researchers by providing detailed reports and analyses of the data which enables insights into drug resistance monitoring, genomic research, outbreak surveillance and epidemiological studies.

Dependencies

- Nextflow
- nf-core

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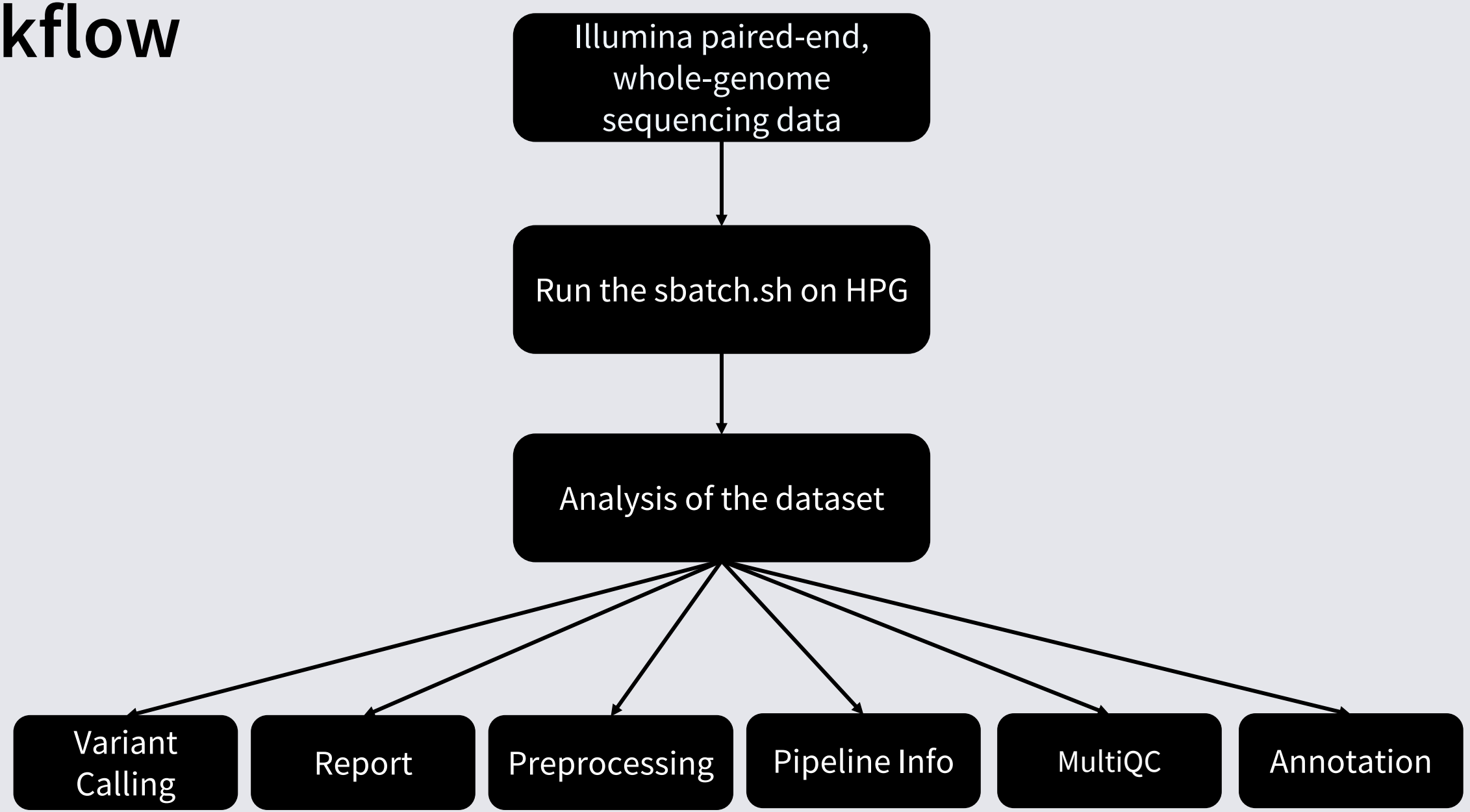
Workflow



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Application

Objective

Use SARS-CoV-19 dataset and analyze using Sarek-Mic Pipeline

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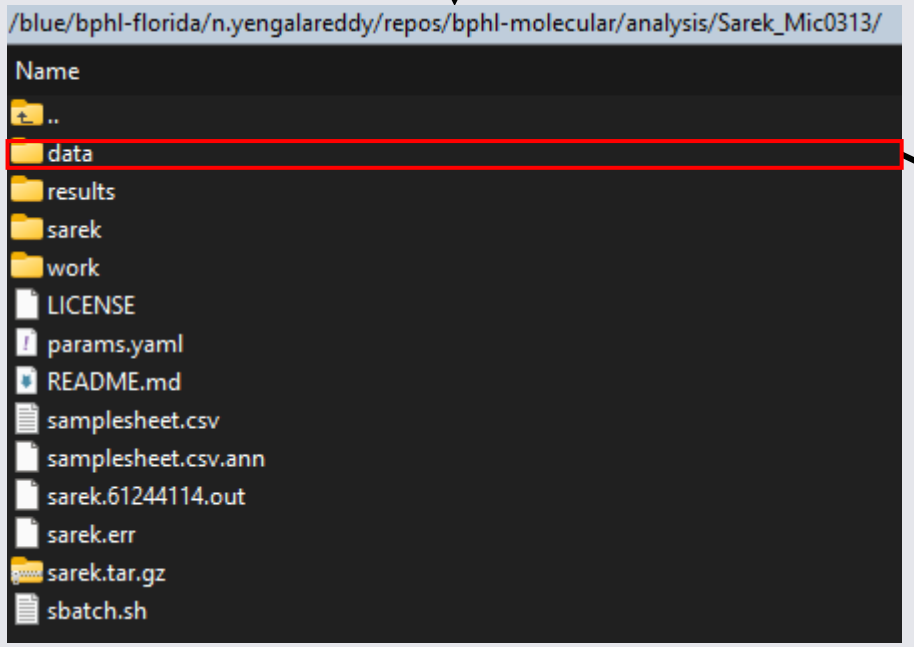
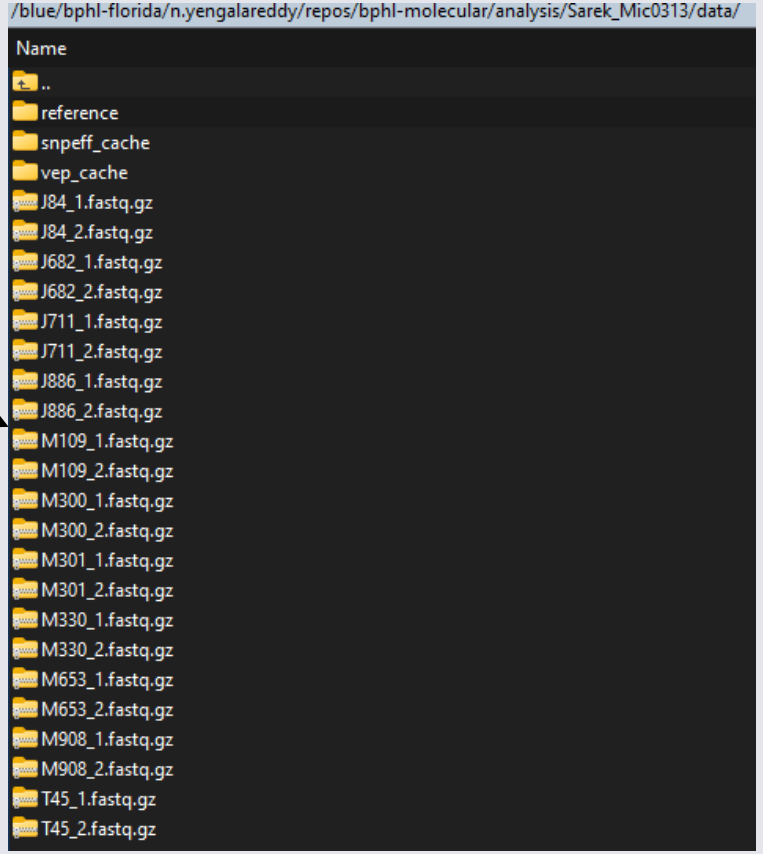
Application Cont.

```
cd /blue/bphl-<state>/<user>/repos/bphl-molecular/
git clone https://github.com/BPHL-Molecular/Sarek_Mic
mkdir analysis/
cd analysis/
cp /blue/bphl-<state>/<user>/repos/bphl-molecular/ Sarek_MIc/
copy .fastq to data directory
```





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Application Cont.

nano params.yaml

/blue/bphl-florida/n.yengalareddy/repos/bphl-molecular/analysis/Sarek_Mic0313/

Name

- ..
- data
- results
- sarek
- work
- LICENSE
- params.yaml
- README.md
- samplesheet.csv
- samplesheet.csv.ann
- sarek.61244114.out
- sarek.err
- sarek.tar.gz
- sbatch.sh

GNU nano 2.9.8 params.yaml

```
input: './samplesheet.csv'
outdir: './results/'
#genome: 'null'
tools: 'mpileup,haplotypecaller,freebayes,strelka,snpeff,vep'
skip_tools: 'baserecalibrator,baserecalibrator_report,dnascope_filter'
#step: 'annotate'
igenomes_ignore: 'true'
snpeff_cache: '/blue/bphl-florida/n.yengalareddy/repos/bphl-molecular/analysis/Sarek_Mic0313/data/snpeff_cache'
snpeff_db: 'MN908947.3'
vep_cache: '/blue/bphl-florida/n.yengalareddy/repos/bphl-molecular/analysis/Sarek_Mic0313/data/vep_cache'
vep_species: 'sars_cov_2'
vep_genome: 'ASM985889v3'
vep_cache_version: '101'

fasta: '/blue/bphl-florida/n.yengalareddy/repos/bphl-molecular/analysis/Sarek_Mic0313/data/reference/covid/nCoV-2019.reference.fasta'
fasta_fai: '/blue/bphl-florida/n.yengalareddy/repos/bphl-molecular/analysis/Sarek_Mic0313/data/reference/covid/nCoV-2019.reference.fasta.fai'
only_paired_variant_calling: 'true'
```

Application Cont.

nano sbatch.sh

/blue/bphl-florida/n.yengalareddy/repos/bphl-molecular/analysis/Sarek_Mic0313/

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GNU nano 2.9.8 sbatch.sh

```
#!/usr/bin/bash
#SBATCH --account=bphl-umbrella
#SBATCH --qos=bphl-umbrella
#SBATCH --job-name=sarek
#SBATCH --ntasks=1
#SBATCH --cpus-per-task=20
#SBATCH --mem=50gb
#SBATCH --time=48:00:00
#SBATCH --output=sarek.%j.out
#SBATCH --error=sarek.err
#SBATCH --mail-user=nikhil.yengala@flhealth.gov
#SBATCH --mail-type=FAIL,END

module load nextflow

APPTAINER_CACHEDIR=./
export APPTAINER_CACHEDIR

#nextflow run nf-core/sarek -profile singularity --input ./samplesheet.csv --outdir ./output --tools 'haplotypecaller,snpeff'
#GRCh38
#nextflow run ./sarek -profile singularity --input ./samplesheet.csv --outdir ./output --tools 'freebayes,snpeff'

#hg38 ucsc from igenomes
#nextflow run ./sarek -profile singularity --input samplesheet.csv --outdir ./ --genome hg38 --tools 'freebayes,snpeff' --skip_tools baserecal

# custom genomes
#nextflow run ./sarek -profile singularity --input samplesheet.csv --outdir ./ --tools 'freebayes' --genome null --igenomes_ignore --fasta ./r
#nextflow run ./sarek -profile hs37d5,singularity --input samplesheet.csv --outdir ./ --tools 'freebayes,snpeff' --skip_tools baserecalibrator
#nextflow run ./sarek -profile singularity --input samplesheet.csv --outdir ./Candida --tools 'freebayes,snpeff' --genome null --igenomes_igno
```

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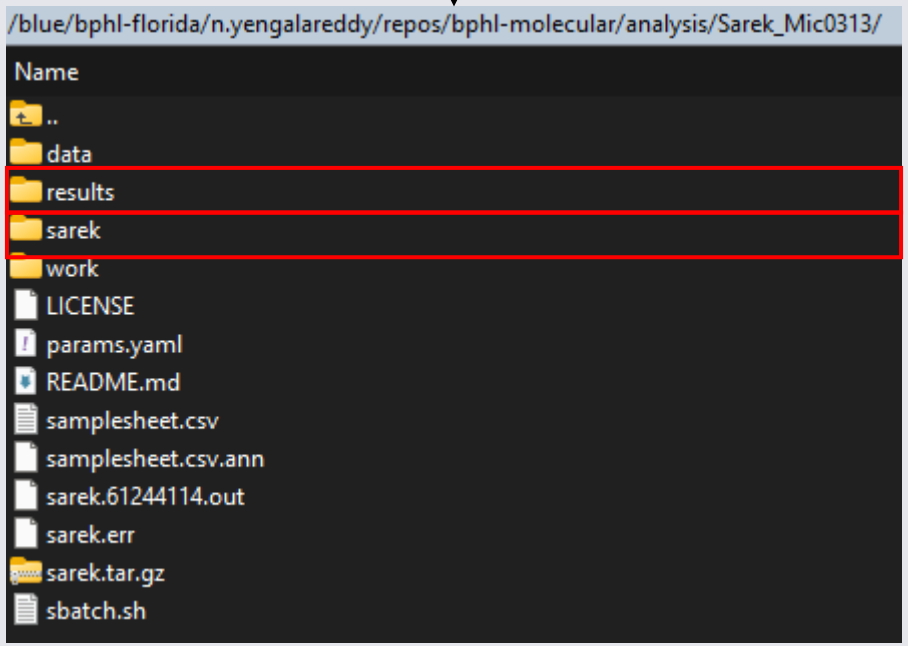
Application Cont.

```
tar xvzf sarek.tar.gz
activate nf-core env
sbatch ./sbatch.sh
```



Application Cont.

/blue/bphl-florida/n.yengalareddy/repos/bphl-molecular/analysis/Sarek_Mic0313/

Name
- ..
- data
- results
- sarek
- work
- LICENSE
- params.yaml
- README.md
- samplesheet.csv
- samplesheet.csv.ann
- sarek.61244114.out
- sarek.err
- sarek.tar.gz
- sbatch.sh

/blue/bphl-florida/n.yengalareddy/repos/bphl-molecular/analysis/Sarek_Mic0313/results/

Name
- ..
- annotation
- build
- csv
- multiqc
- pipeline_info
- preprocessing
- reference
- reports
- variant_calling

/.../n.yengalareddy/repos/bphl-molecular/analysis/Sarek_Mic0313/results/annotation/

Name
- ..
- bcftools
- freebayes
- strelka

/.../repos/bphl-molecular/analysis/Sarek_Mic0313/results/annotation/bcftools/

Name
- ..
- J84
- J682
- J711
- J886
- M109
- M300
- M301
- M330
- M653
- M908
- T45

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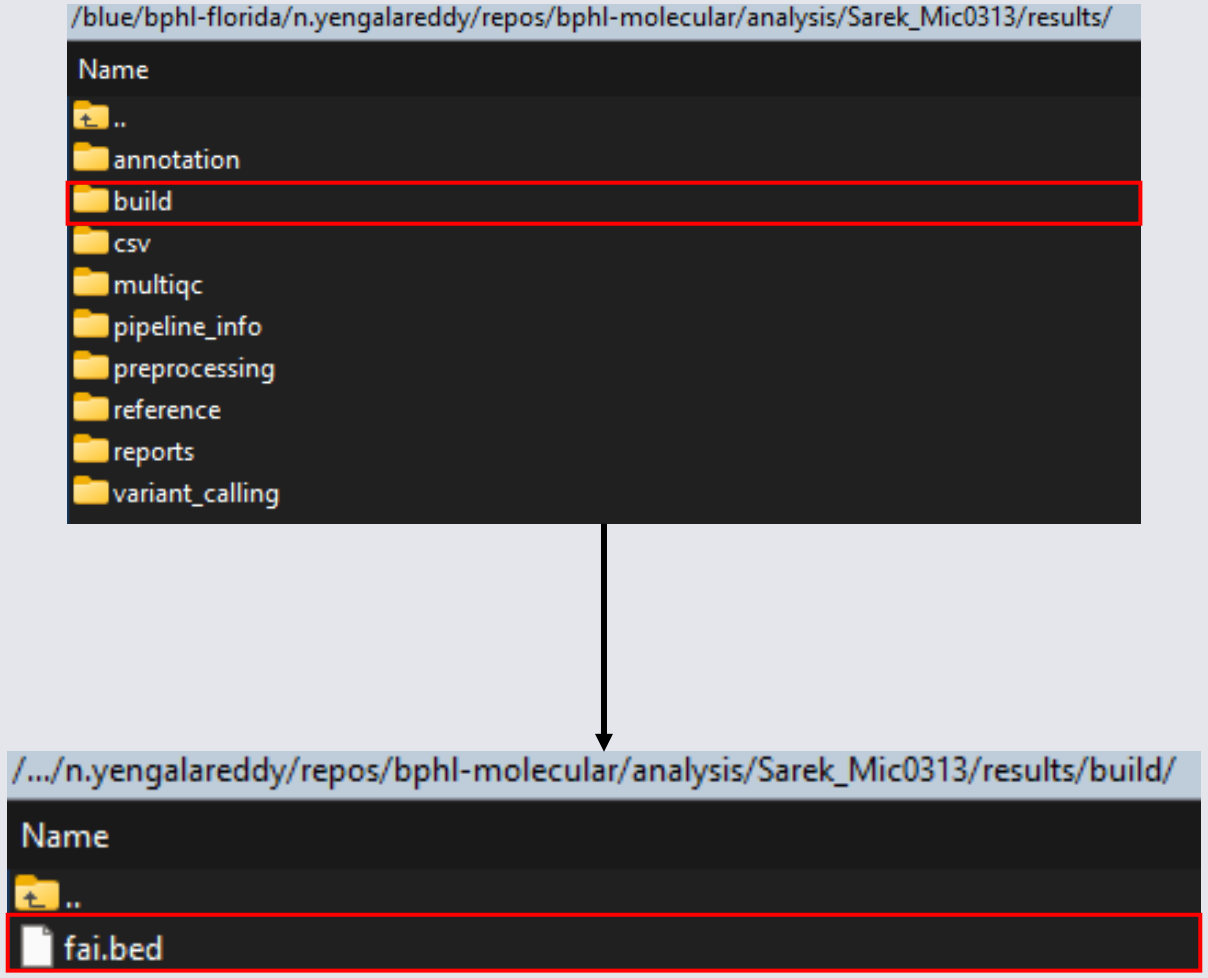
Application Cont.



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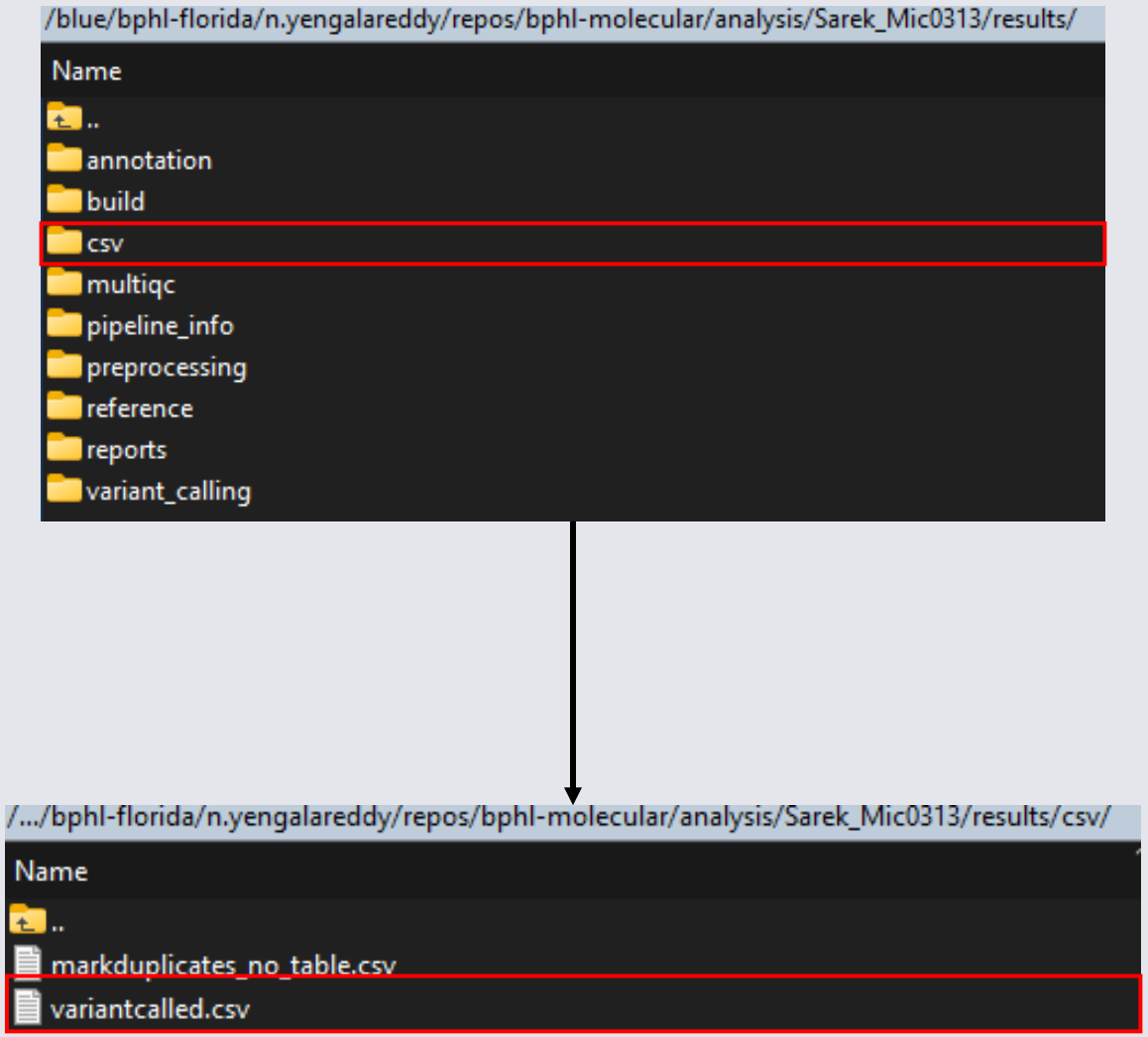
Application Cont.



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Application Cont.

/blue/bphl-florida/n.yengalareddy/repos/bphl-molecular/analysis/Sarek_Mic0313/results/

Name

- ..
- annotation
- build
- csv
- multiqc
- pipeline_info
- preprocessing
- reference
- reports
- variant_calling

/.../n.yengalareddy/repos/bphl-molecular/analysis/Sarek_Mic0313/results/multiqc/

Name

- ..
- multiqc_data
- multiqc_plots
- multiqc_report.html

/.../repos/bphl-molecular/analysis/Sarek_Mic0313/results/multiqc/multiqc_data/

Name

- ..
- mosdepth_cov_dist.txt
- mosdepth_cumcov_dist.txt
- mosdepth_perchrom.txt
- multiqc.log
- multiqc_bcftools_stats.txt
- multiqc_citations.txt
- multiqc_data.json
- multiqc_fastp.txt
- multiqc_fastqc.txt
- multiqc_general_stats.txt
- multiqc_picard_dups.txt
- multiqc_samtools_stats.txt
- multiqc_snpeff.txt
- multiqc_software_versions.txt
- multiqc_sources.txt
- picard_histogram.txt
- picard_histogram_1.txt
- picard_histogram_2.txt
- vcftools_tstv_by_count.txt
- vcftools_tstv_by_qual.txt
- vep.txt

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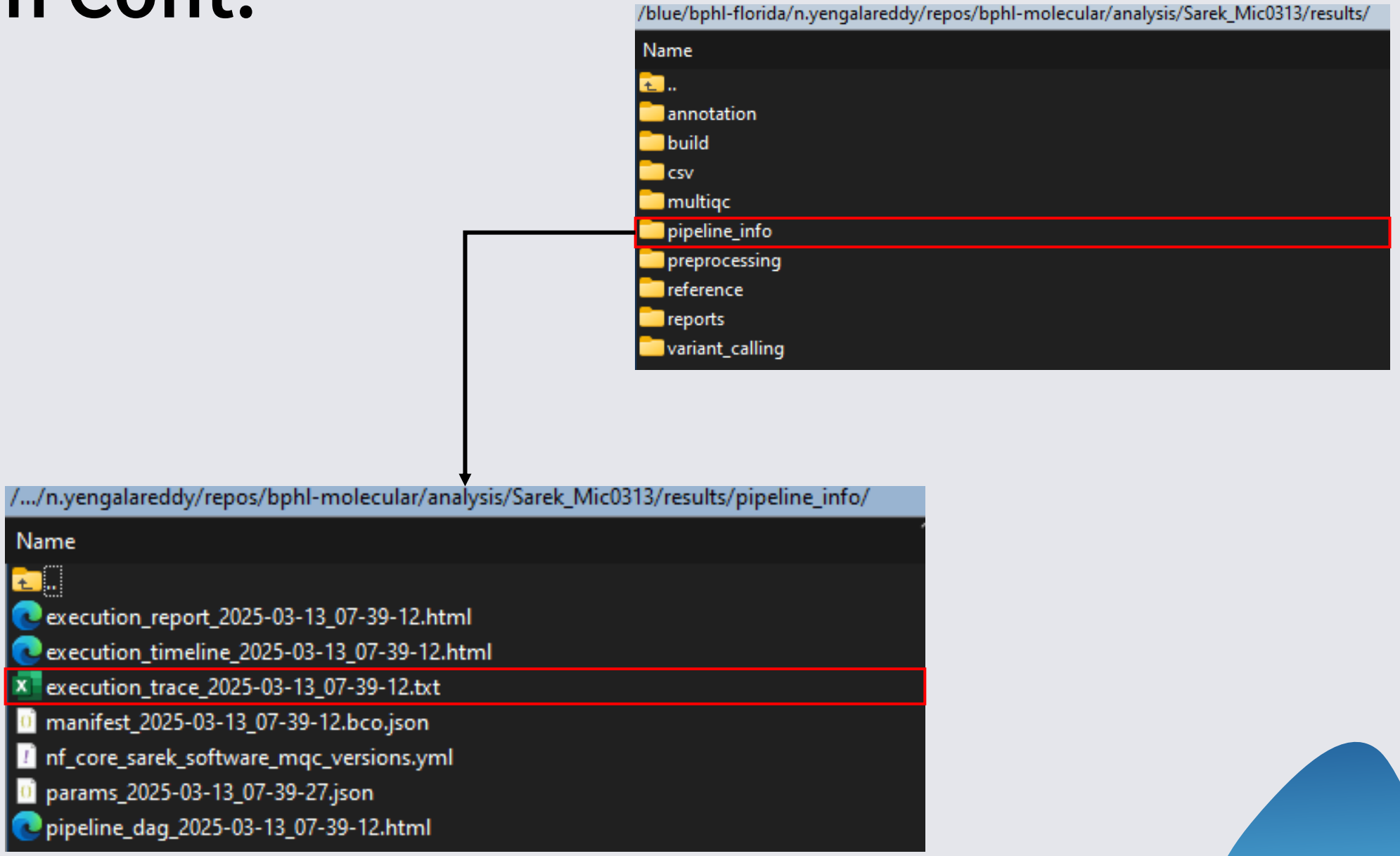
Application Cont.



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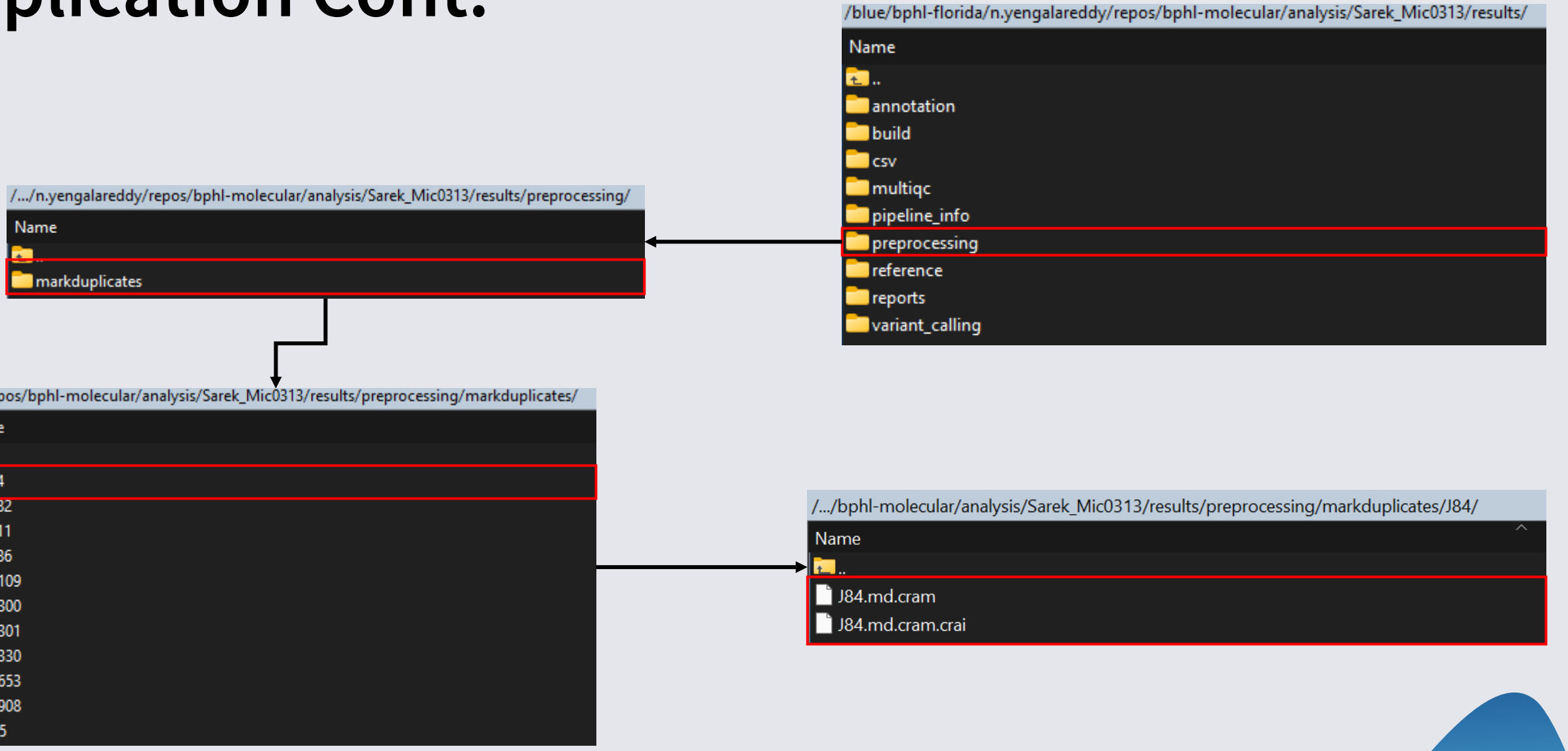
Application Cont.



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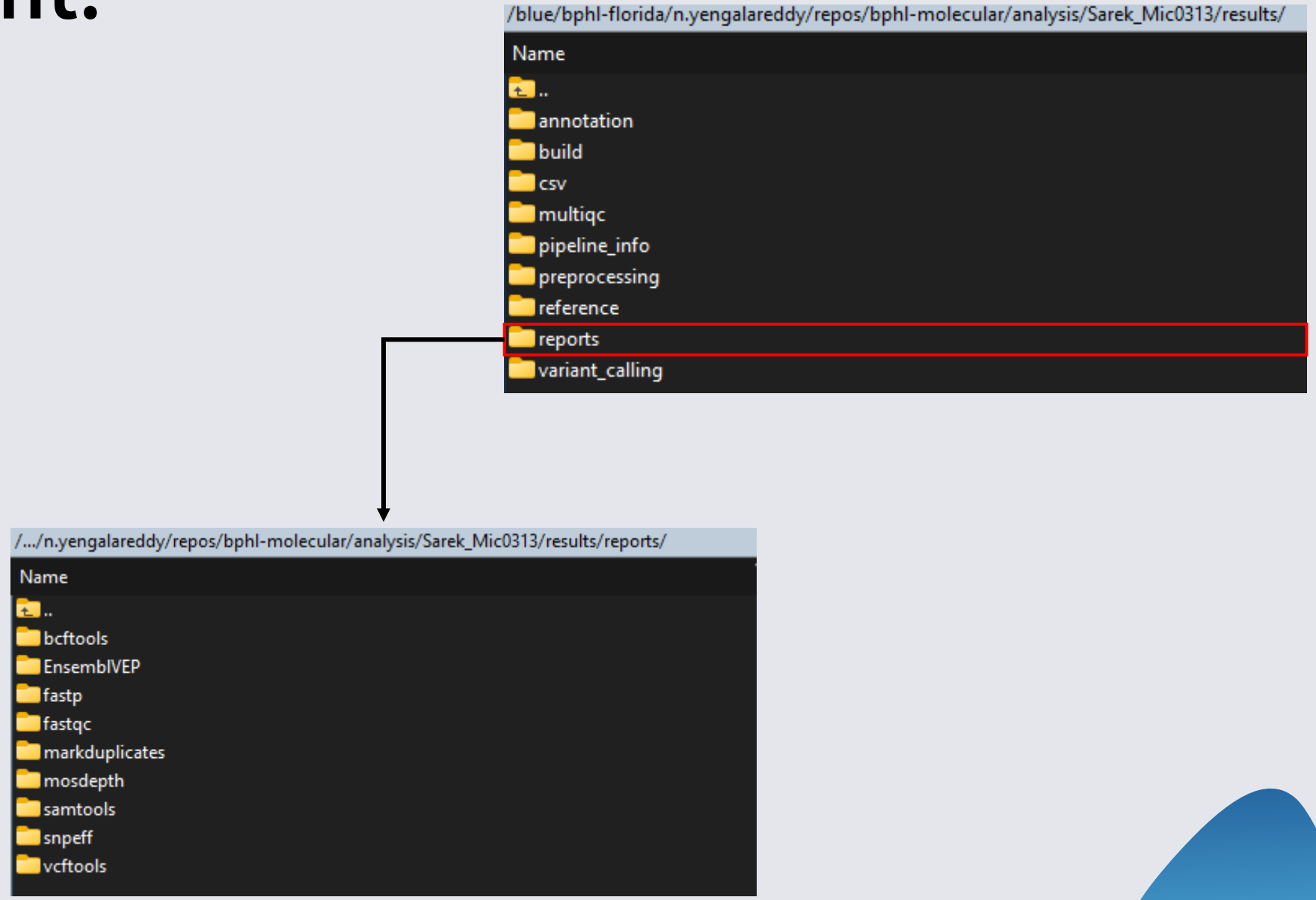

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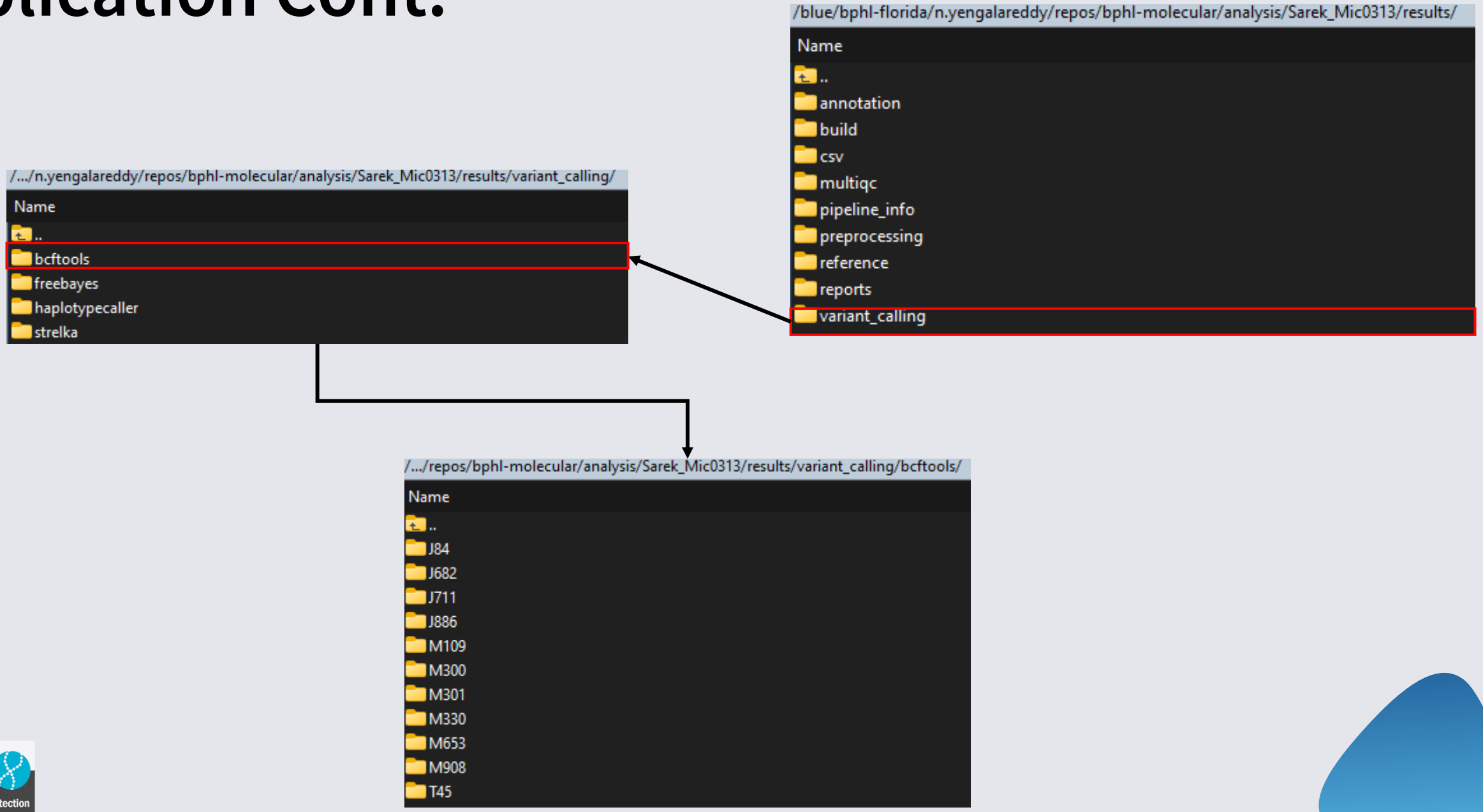

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Conclusion

- Fundamentals of Sarek_Mic
- Installation and setup of Sarek_Mic in HPG
- Successfully executed job query for Sarek_Mic
- Generated output files

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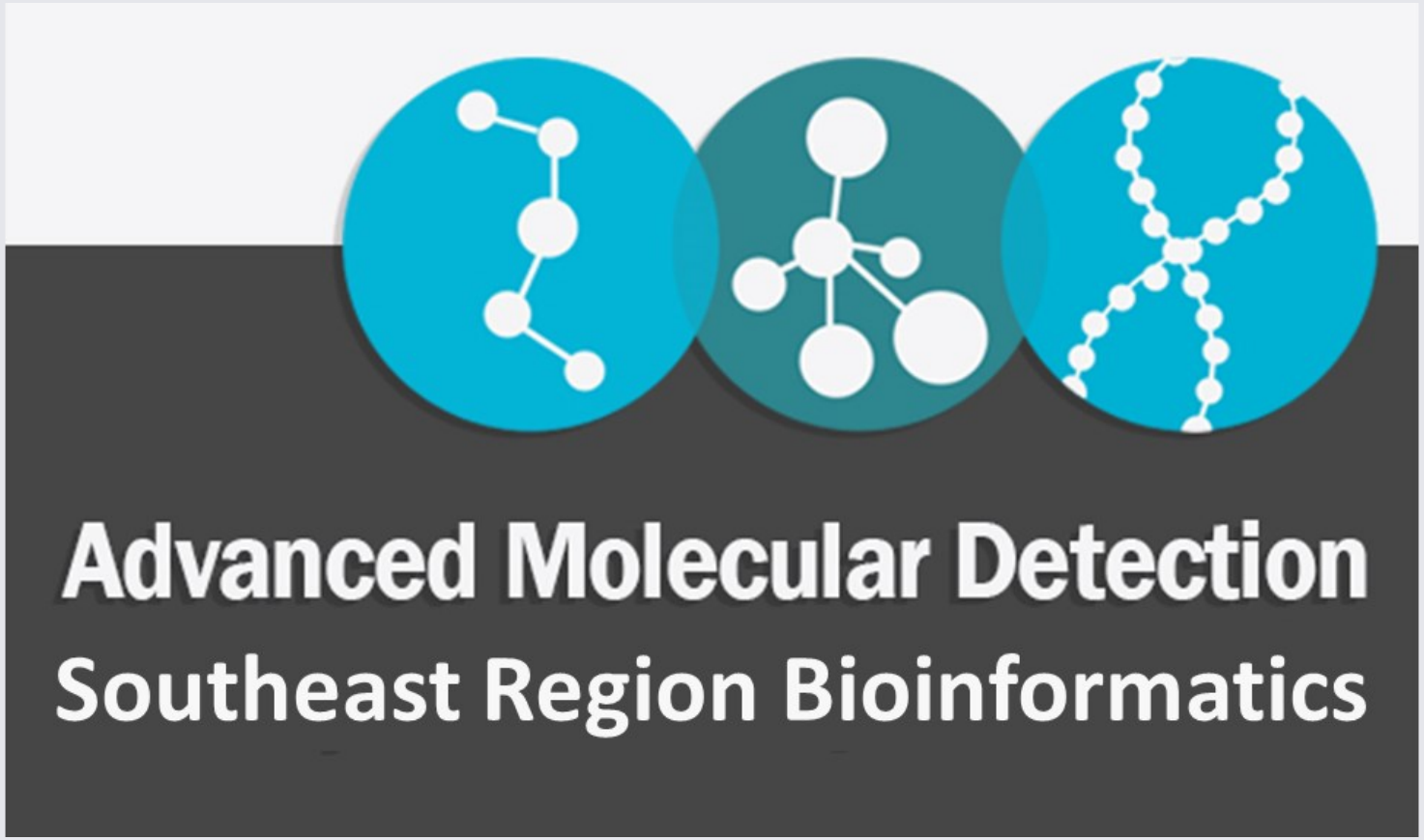

Questions?

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