Bioinformatics Quality Management System

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Objectives

- Describe quality management system (QMS) components
  - Reproducibility
  - Version control
  - Documentation
- Apply QMS principles to NGS workflows
  - Pre-analytical
  - Analytical
- Common troubleshooting

Reproducibility

- Same input + same methods = same results
  - Core principle of computational (all) science

- Record software versions
  - Aligners, samtools, variant callers, workflow managers
- Track changes with version control
  - Git for scripts and pipeline development
- Document parameters and references
  - Primer schemes
  - Quality thresholds

- Identify test data to run through new versions and pipelines

- Use workflow managers
  - Snakemake, Nextflow for structured, repeatable runs
- Leverage containers
  - Docker or Singularity ensure consistent environments

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Documentation

- Explains how tools and pipelines work
  - Inputs, outputs, parameters, and assumptions
- Enables reproducibility
  - Others (and future you) can rerun the same analysis
- Reduces errors and misuse
  - Clear defaults and examples prevent incorrect runs
- Essential for collaboration
  - Shared understanding across labs, teams, and institutions

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Best Practices in writing documentation

- README.md files
  - Overview, requirements, and basic usage
- Usage examples & command snippets
  - Copy-pasteable starting points
- Parameter descriptions
  - What flags do, expected values, and defaults
- Workflow diagrams
  - Visualize steps, inputs, and outputs

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Best Practices in writing documentation

- Document data formats
    - FASTQ, FASTA, BAM, reference versions
- Record software versions
    - Tools, containers, workflow managers
- Include example datasets
    - Small test cases to validate setup

Best Practices in using documentation

- Start with the README
  - Understand the purpose, inputs, outputs, and limitations
- Check versions and dates
  - Ensure the documentation matches the tool or pipeline version you're using
- Follow the example first
  - Run the provided test or example before real data
- Read parameter descriptions carefully
  - Defaults may not match your experiment
- Verify input formats
  - FASTQ, FASTA, BAM, sample sheets, reference builds
- Look for expected outputs
  - Confirm files and directories match the documented results

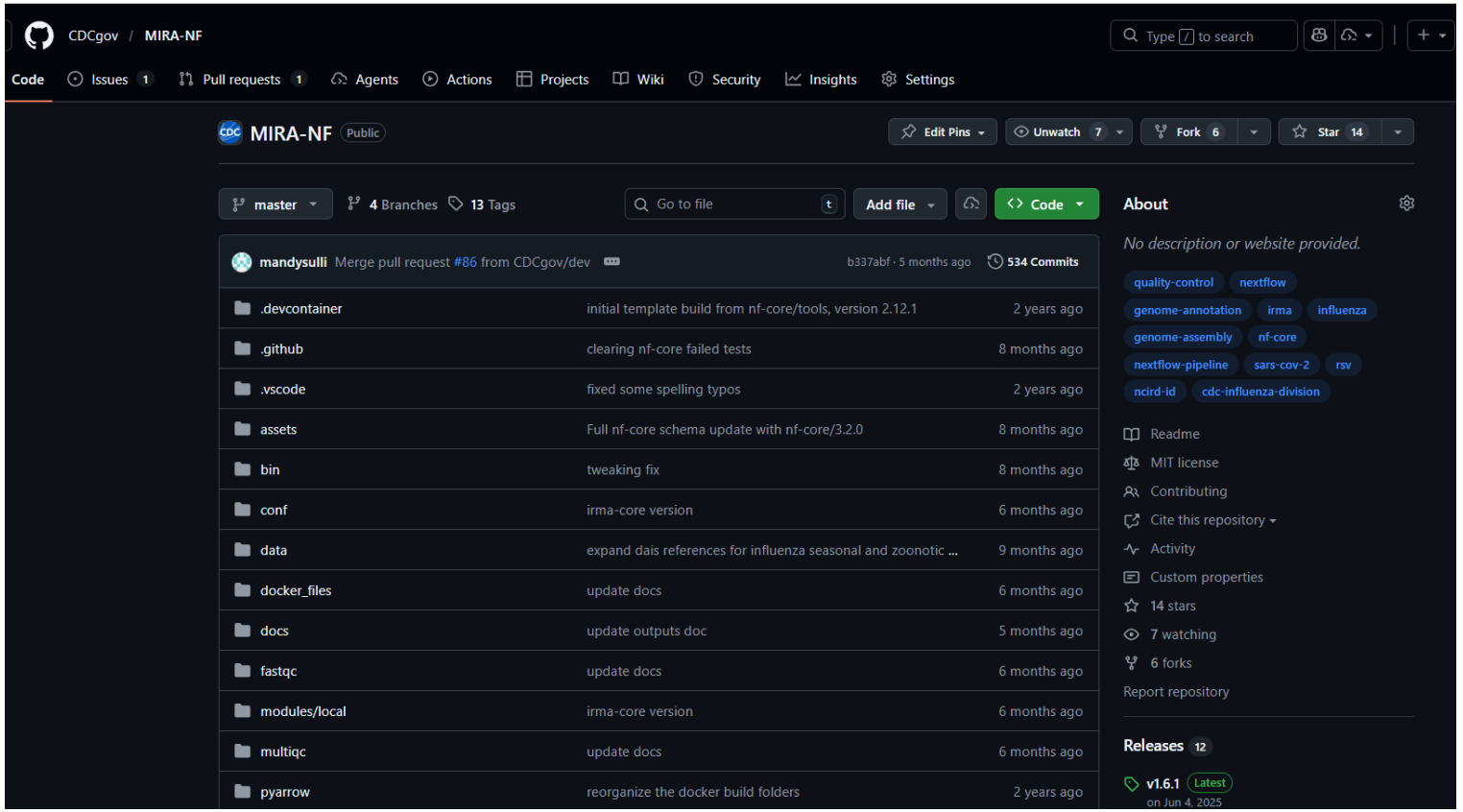
Git

- Git is a version control system
  - Tracks changes to files over time
- Designed for collaboration
  - Multiple people can work on the same codebase
  - Think of it like a shared document (Microsoft office, Google docs)
- Keeps a history of your work
  - See what changed, when, why, and by whom
- Common in bioinformatics
  - Pipelines, scripts, documentation, and workflows

- Gitlab and Github most used

Core Git concepts

- Repository (repo)
  - A project tracked by Git
- Commit
  - A saved snapshot of changes with a message
- Branch
  - A parallel version of the code for development or testing
- Remote repository
  - Shared copy on platforms like GitHub or GitLab




```
qgx6@rosalind02:MIRA-NF$ git status
On branch dev
Your branch is up to date with 'origin/dev'.

Untracked files:
  (use "git add <file>..." to include in what will be committed)
        .MIRA_nextflow.sh

nothing added to commit but untracked files present (use "git add" to track)
```

Git

- Common Git Commands (User Level)
- Create or get a repository
  - git init — start a repo
  - git clone <repo> — copy an existing repo
- Track and save changes
  - git status — see changes
  - git add <file> — stage changes
  - git commit -m "message" — save snapshot
- Sync with others
  - git pull — get updates
  - git push — share your changes