1. Accounts

Select either AWS or Local Cluster

If AWS

Provide your AWS login credentials

A session ID will be generated for future logins

If Local Cluster

Provide your login credentials to your local cluster

2. Select Genomes

Select either an EBS volume (only with AWS accounts), user defined, or 1000 Genomes

Click “Load list” to load the data

Choose the .bam files to use

Choose a reference genome

Select if you want a copy of all sorted .bam and .bai created to storage directory

3. Configure Tools

Execution Profiles

New

Select a Run mode (germline, somatic, de novo/ family trio)

Select Paramaters (these can be tuned further in each tool)

Existing

Upload from file

Select Genomic Regions

No selection

Standard chromosome1-22, X, Y

Standard chromosomes plus contigs MT, GI, GL, NC

User defined either manual entry or file upload

Click the “Apply Profile” button to apply above entries

Customization

For each tool switched on by the run mode selected, fine tune any parameters. If necessary, turn off any tools switched on or turn on any tools not automatically selected.

3. Submit

If AWS

Select an AWS computing resource instance

Select a storage bucket to put results

If Local Cluster

Select a local computing resource

Choose a working directory to run jobs

Select submit action (create script vs. execute script)

Specify any additional comments (optional)

Double Check

Click the “Preview” button to check execution profile

Click the “Validate” button to preview and check user settings

Click “Submit”

4. Results

Check out the results by navigating to the location displayed (for both AWS and Local Cluster)

Enjoy!

5. Options

Additional options and accounts management may be configured for AWS