

Supporting validated community developed variant calling analyses

Brad Chapman

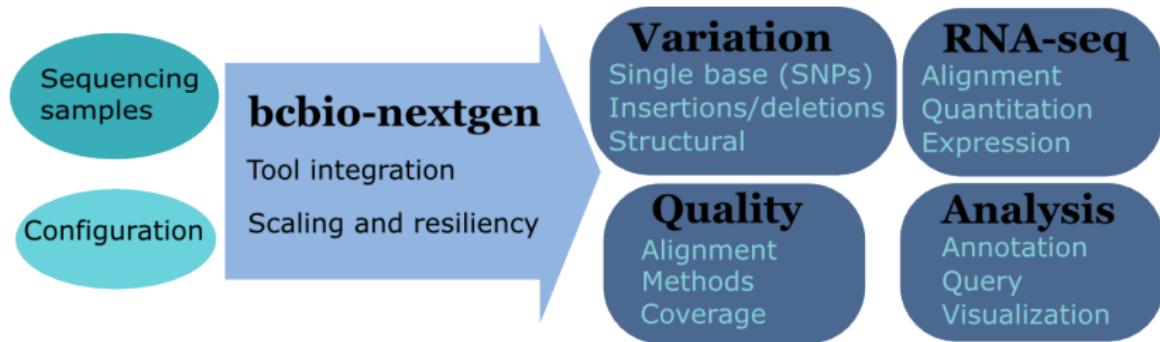
Bioinformatics Core, Harvard School of Public Health

<https://github.com/chapmanb/bcbio-nextgen>

<http://j.mp/bcbiolinks>

12 July 2014

Overview



<https://github.com/chapmanb/bcbio-nextgen>

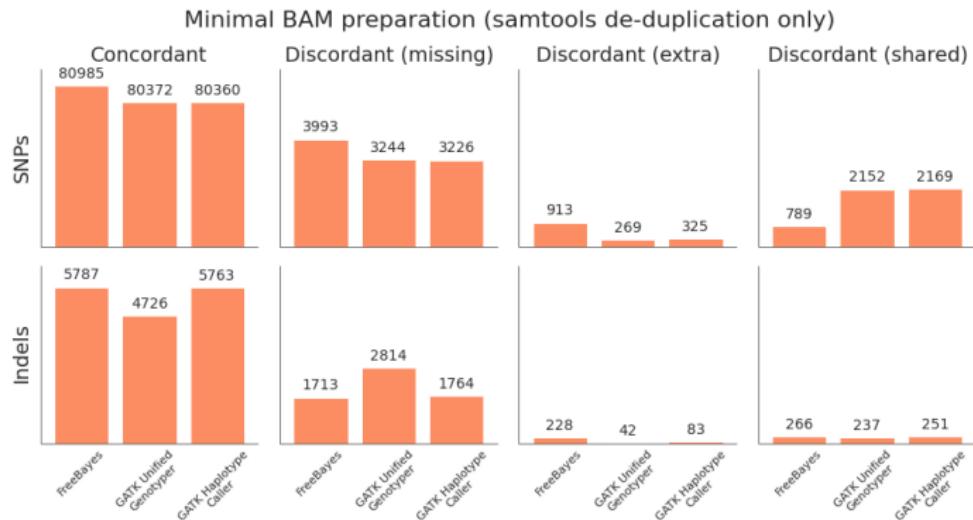
Uses

- Aligners: bwa-mem, novoalign, bowtie2
- Variantion: FreeBayes, GATK, MuTecT, Scalpel, SnpEff, VEP, GEMINI, Lumpy, Delly
- RNA-seq: Tophat, STAR, cufflinks, HTSeq
- Quality control: fastqc, bamtools, RNA-SeQC
- Manipulation: bedtools, bcftools, biobambam, sambamba, samblaster, samtools, vcflib

Provides

- Community – collected set of expertise
- Tool integration
- Validation – outputs + automated evaluation
- Installation of tools and data
- Scaling

Validation > Replication



Genome in a Bottle: <http://www.genomeinabottle.org/>

ICGC-TCGA DREAM: <https://www.synapse.org/#!Synapse:syn312572>

SMaSH: <http://smash.cs.berkeley.edu/>

Make installation easy



John Davey
@johnomics



Following

The trepidation of opening an INSTALL file.

"Please say ./configure; make; make
install... please say ./configure; make; make
install..."

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Automated Install

We made it easy to install a large number of biological tools.
Good or bad idea?

Need a consistent support environment

Code 18

Issues 104

Installation

We've found 104 issues

States

Closed	96
Open	8

Search all of GitHub

Installation can fail if pypi is blocked
Opened by lbeltrame 2 days ago

Mac OS 10.9 installation error
Opened by alartin on Apr 13 2 comments

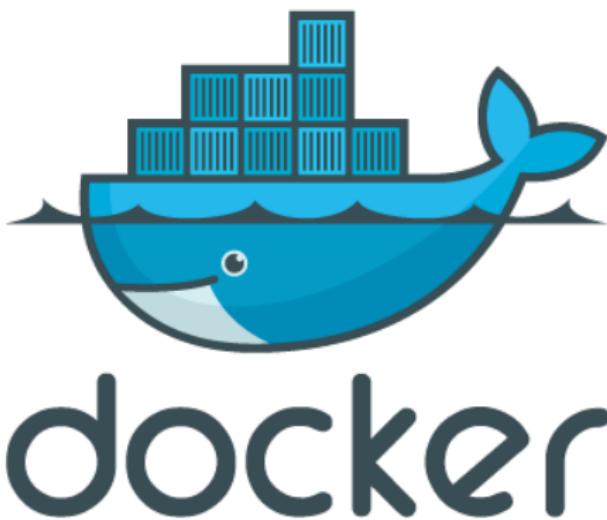
Update installation.rst
add --data to dbnsfp download
Opened by tanglingfung 26 days ago 1 comment

SHA256 mismatch for platypus-variant in installation
Hi, I encountered an error when installing the latest version of bcbio-nextgen on Ubuntu
installation halted with a SHA256 mismatch error when it was installing platypus-varian
Opened by kennethban 3 days ago 2 comments

Installation in arch
Opened by kspham on Jun 12 1 comment

Navigation icons: back, forward, search, etc.

Docker lightweight containers



<http://docker.io>

Docker benefits

- Fully isolated
- Reproducible – store full environment with analysis (1Gb)
- Improved installation – single download + data

bcbio with Docker

- External Python wrapper
 - Installation
 - Start and run containers
 - Mount external data into containers
 - Parallelize
- All analysis tools inside Docker

<https://github.com/chapmanb/bcbio-nextgen-vm>
<http://j.mp/bcbiodocker>

Docker image automation

```
$ ansible-playbook bcbio_vm_aws.yml  
$ docker import \  
https://s3.amazonaws.com/bcbio_nextgen/ \  
    bcbio-nextgen-docker-image.gz \  
chapmanb/bcbio-nextgen-devel
```

<http://www.ansible.com>

[https://github.com/chapmanb/bcbio-nextgen-vm/tree/
master/ansible](https://github.com/chapmanb/bcbio-nextgen-vm/tree/master/ansible)

Docker HPC parallelization

bcbio-nextgen-vm
bcbio-nextgen
(workflow and parallel)
IPython parallel

Cluster scheduler
(SLURM, Torque,
SGE, LSF)

Machine 1

Docker Container
bcbio-nextgen
(run tools)
external tools
(bwa, freebayes...)

Machine 2

Docker Container
bcbio-nextgen
(run tools)
external tools
(bwa, freebayes...)

<http://ipython.org/ipython-doc/dev/parallel/index.html>
<https://github.com/roryk/ipython-cluster-helper>

Consistent scaling environment



Amazon challenges

- Cost – spot instances
- Disk – local scratch, no EBS
- Organization – no shared filesystems,
S3 push/pull
- Data – reconstitute on minimal machines
- Security – encryption at rest

Amazon approaches

- Clusterk <http://clusterk.com/>
- Arvados <http://arvados.org/>
- Galaxy <http://usegalaxy.org/>

Summary

- Community developed variant calling analyses
<https://github.com/chapmanb/bcbio-nextgen>
- Docker: consistent install environment
- Automation: reproducible, understandable builds
- Need for a consistent scaling environment