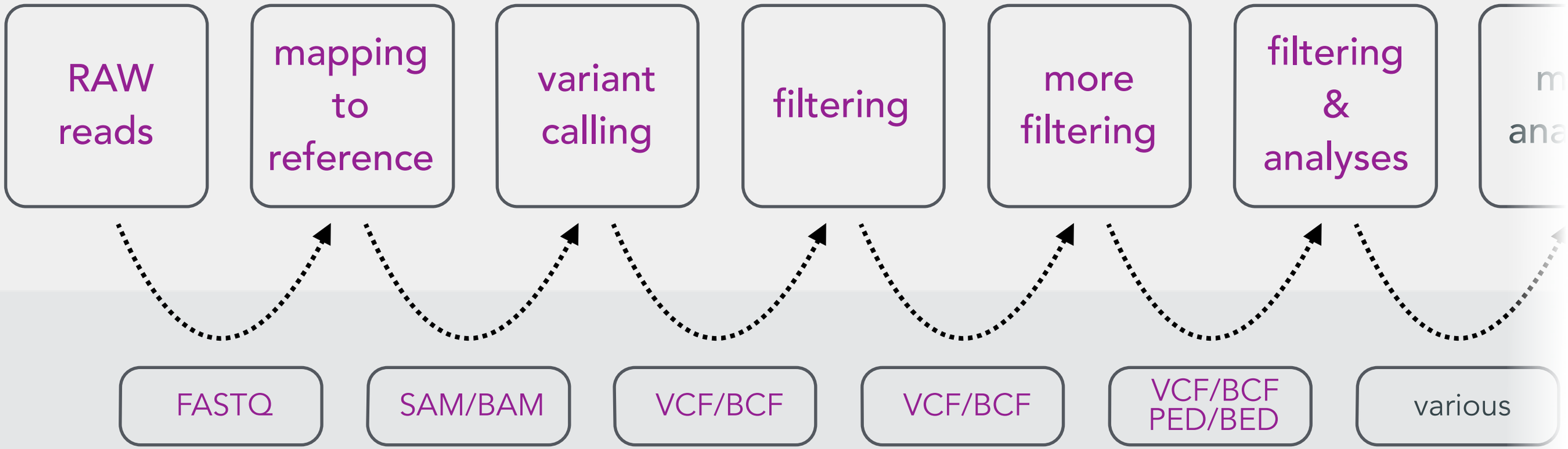
FileFormats.VCFtools.PLINK

Pipeline



File format

Pipeline

RAW reads

mapping to reference

variant calling

filtering

more filtering

filtering & analyses

m ana

FASTQ

SAM/BAM

VCF/BCF

VCF/BCF

VCF/BCF PED/BED

various

File format

now!

tonight

tomorrow

Name

bcftools — utilities for variant calling and manipulating VCFs and BCFs.

Synopsis

bcftools [--version|--version-only] [--help] [*COMMAND*] [*OPTIONS*]

DESCRIPTION

BCFtools is a set of utilities that manipulate variant calls in the Variant Call Format (VCF) and its binary counterpart BCF. All commands work transparently with both VCFs and BCFs, both uncompressed and BGZF-compressed.

Most commands accept VCF, bgzipped VCF and BCF with filetype detected automatically even when streaming from a pipe. Indexed VCF and BCF will work in all situations. Un-indexed VCF and BCF and streams will work in most, but not all situations. In general, whenever multiple VCFs are read simultaneously, they must be indexed and therefore also compressed.

BCFtools is designed to work on a stream. It regards an input file "-" as the standard input (stdin) and outputs to the standard output (stdout). Several commands can thus be combined with Unix pipes.

VERSION

This manual page was last updated **2015-12-15 14:02 GMT** and refers to bcftools git version **1.2-191-g6737c5c+**.

BCF1

The BCF1 format output by versions of samtools <= 0.1.19 is **not** compatible with this version of bcftools. To read BCF1 files one can use the view command from old versions of bcftools packaged with samtools versions <= 0.1.19 to convert to VCF, which can then be read by this version of bcftools.

```
samtools-0.1.19/bcftools/bcftools view file.bcf1 | bcftools view
```

VARIANT CALLING

See *bcftools call* for variant calling from the output of the *samtools mpileup* command. In versions of samtools <= 0.1.19 calling was done with *bcftools view*. Users are now required to choose between the old samtools calling model (*-c/--consensus-caller*) and

VCFtools

A set of tools written in Perl and C++ for working with VCF files.

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Welcome to VCFtools

VCFtools is a program package designed for working with VCF files, such as those generated by the 1000 Genomes Project. The aim of VCFtools is to provide easily accessible methods for working with complex genetic variation data in the form of VCF files.

This toolset can be used to perform the following operations on VCF files:

- Filter out specific variants
- Compare files
- Summarize variants
- Convert to different file types
- Validate and merge files
- Create intersections and subsets of variants

VCFtools consists of two parts, a **perl module** and a **binary executable**. The perl module is a general Perl API for manipulating VCF files, whereas the binary executable provides general analysis routines.

Download

To obtain VCFtools, please visit the downloads page.

Variant call format specification

VCFtools is compatible with VCF versions 4.0, 4.1 and 4.2. For more information regarding the VCF format, please visit the VCF specification page.

Contact

plink...

*Last original PLINK release is **v1.07** (10-Oct-2009); **PLINK 1.9** is now available for beta-testing*

Whole genome association analysis toolset

New (15-May-2014): PLINK 1.9 is now available for beta-testing!

PLINK is a free, open-source whole genome association analysis toolset, designed to perform a range of basic, large-scale analyses in a computationally efficient manner.

The focus of **PLINK** is purely on *analysis* of genotype/phenotype data, so there is no support for steps prior to this (e.g. study design and planning, generating genotype or CNV calls from raw data). Through integration with gPLINK and Haploview, there is some support for the subsequent visualization, annotation and storage of results.

PLINK (one syllable) is being developed by Shaun Purcell at the Center for Human Genetic Research (CHGR), Massachusetts General Hospital (MGH), and the Broad Institute of Harvard & MIT, with the support of others.

Quick links

PLINK tutorial

gPLINK

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Resources

FAQs | PDF

Citing PLINK

Bugs, questions?

New in 1.07: meta-analysis, result annotation and analysis of dosage data.

Data management

- Read data in a variety of formats
- Recode and reorder files
- Merge two or more files
- Extracts subsets (SNPs or individuals)
- Flip strand of SNPs
- Compress data in a binary file format

Summary statistics for quality control

PLINK 1.90 beta

This is a comprehensive update to Shaun Purcell's PLINK command-line program, developed by Christopher Chang with support from the NIH-NIDDK's Laboratory of Biological Modeling, the Purcell Lab at Mount Sinai School of Medicine, and others. (What's new?) (Credits.) (Methods paper.)

Binary downloads

	Build		
Operating system[1]	**Stable (beta 3.29, 24 Dec)**	Development (23 Jan)	Old[2] (v1.07)
Linux 64-bit	**download**	download	download
Linux 32-bit	**download**	download	download
OS X (64-bit)	**download**	download	download
Windows 64-bit	**download**	download	download
Windows 32-bit	**download**	download	

1: Solaris is no longer explicitly supported, but it should be able to run the Linux binaries.
2: These are just mirrors of the binaries posted at http://pngu.mgh.harvard.edu/~purcell/plink/download.shtml.

Source code, compilation instructions, and the like are on the developer page.

The following documented PLINK 1.07 flags are not supported by 1.90 beta 3:

- --qual-geno-scores[3]
- --segment[4]
- --dfam
- --p2, --genedrop
- --hap, --hap-window, --hap-snps[5]
- --proxy-assoc, --proxy-impute[5]
- --cnv-list, --cfile, --gfile

http://evomics.org/learning/population-and-speciation-genomics/fileformats-vcftools-plink/