SIMdrom Documentation

Release

Max Schubach

1	API Documentation	1
2	Indices and tables	21

CHAPTER 1

API Documentation

The friendly people at javadoc.io host our API documentation:

simdrom-core https://javadoc.io/doc/de.charite.compbio/simdrom-core/0.0/

simdrom-cli https://javadoc.io/doc/de.charite.compbio/simdrom-cli/0.0/

Quickstart

This short How-To guides you from downloading the SIMdrom program and to simualte a new individual.

- 1. Download the current stable release from our GitHub project by clicking here.
- 2. Extract the ZIP archive.
 - you should find file called ${\tt simdrom-cli-|version|}$. jar in the ZIP
 - you should also find a file small.vcf file in the folder examples

Installation

Pre-built Binaries

Note: This is the recommended way of installing for normal users.

Pre-built binaries are available from Maven Central. Download sidmrom-cli-0.0 .jar from here.

Install from Source

Note: You only need to install from source if you want to develop SIMdrom in Java yourself.

There are two options of installing SIMdrom. The recommended way for most users is to download a prebuilt binary and is well-described in the *Quickstart* section. This section describes how to build SIMdrom from scratch.

Prequisites

For building SIMdrom, you will need

- 1. Java JDK 8 or higher for compiling Jannovar,
- 2. Maven 3 for building Jannovar, and
- 3. Git for getting the sources.

Git Checkout

In this tutorial, we will download the SIMdrom sources and build them in ~/Development/SIMdrom.

```
~ # mkdir -p ~/SIMdrom
~ # cd ~/SIMdrom
SIMdrom # git clone https://github.com/visze/simdrom.git simdrom
SIMdrom # cd simdrom
```

Maven Proxy Settings

If you are behind a proxy, you will get problems with Maven downloading dependencies. If you run into problems, make sure to also delete $\sim/.m2/repository$. Then, execute the following commands to fill $\sim/.m2/settings.xml$.

Building

You can build SIMdrom using mvn package. This will automatically download all dependencies, build SIMdrom, and run all tests.

```
simdrom # mvn package
```

In case that you have non-compiling test, you can use the -DskipTests=true parameter for skipping them.

```
simdrom # mvn install -DskipTests=true
```

Creating Eclipse Projects

Maven can be used to generate Eclipse projects that can be imported by the Eclipse IDE. This can be done calling mvn eclipse:eclipse command after calling mvn install:

```
simdrom # mvn install
simdrom # mvn eclipse:eclipse
```

Frequently Asked Questions

Where are the questions?

Right now, there are no frequently asked questions.

Contributing

Contributions are welcome, and they are greatly appreciated! Every little bit helps, and credit will always be given.

You can contribute in many ways:

Types of Contributions

Report Bugs

Report bugs at https://github.com/visze/simdrom/issues

If you are reporting a bug, please include:

- Your operating system name and version.
- Any details about your local setup that might be helpful in troubleshooting.
- Detailed steps to reproduce the bug.

Fix Bugs

Look through the Github issues for bugs. If you want to start working on a bug then please write short message on the issue tracker to prevent duplicate work.

Implement Features

Look through the Github issues for features. If you want to start working on an issue then please write short message on the issue tracker to prevent duplicate work.

Write Documentation

SIMdrom could always use more documentation, whether as part of the official vcfpy docs, in docstrings, or even on the web in blog posts, articles, and such.

SIMdrom uses Sphinx for the user manual (that you are currently reading). See *doc_guidelines* on how the documentation reStructuredText is used. See *doc_setup* on creating a local setup for building the documentation.

Submit Feedback

The best way to send feedback is to file an issue at https://github.com/charite/jannovar/issues

If you are proposing a feature:

- Explain in detail how it would work.
- Keep the scope as narrow as possible, to make it easier to implement.
- Remember that this is a volunteer-driven project, and that contributions are welcome:)

Documentation Guidelines

For the documentation, please adhere to the following guidelines:

- Put each sentence on its own line, this makes tracking changes through Git SCM easier.
- Provide hyperlink targets, at least for the first two section levels.
- Use the section structure from below.

```
.. heading_1:
========
Heading 1
=======

.. heading_2:
------
Heading 2
------
.. heading_3:
Heading 3
=======

.. heading_4:
Heading 4
------
.. heading_5:
Heading 5
```

```
.. heading_6:
Heading 6
::::::::
```

Documentation Setup

For building the documentation, you have to install the Python program Sphinx. This is best done in a virtual environment. The following assumes you have a working Python 3 setup.

Use the following steps for installing Sphinx and the dependencies for building the SIMdrom documentation:

```
$ cd simdrom/manual
$ virtualenv -p python3 .venv
$ source .venv/bin/activate
$ pip install --upgrade -r requirements.txt
```

Use the following for building the documentation. The first two lines is only required for loading the virtualenv. Afterwards, you can always use make html for building.

```
$ cd simdrom/manual
$ source .venv/bin/activate
$ make html # rebuild for changed files only
$ make clean && make html # force rebuild
```

Get Started!

Ready to contribute? First, create your Java/Documentation development setup as described in *in-stall_from_sourceldoc_setup*.

- 1. Fork the *simdrom* repo on GitHub.
- 2. Clone your fork locally:

```
$ git clone git@github.com:your_name_here/simdrom.git
```

3. Create a branch for local development:

```
$ git checkout -b name-of-your-bugfix-or-feature
```

Now you can make your changes locally.

5. When you're done making your changes, make sure that the build runs through. For Java:

\$ mvn package

For documentation:

\$ make clean && make html

6. Commit your changes and push your branch to GitHub:

1.4. Contributing 5

```
$ git add .
$ git commit -m "Your detailed description of your changes."
$ git push origin name-of-your-bugfix-or-feature
```

7. Submit a pull request through the GitHub website.

Pull Request Guidelines

Before you submit a pull request, check that it meets these guidelines:

- 1. The pull request should include tests.
- 2. If the pull request adds functionality, the docs should be updated.
- 3. Describe your changes in the CHANGELOG.md file.

Authors

in alphabetical order

- Sofia Ånäs
- · Max Schubach

History

```
# SIMdrom Changelog
## development
### simdrom-cli
* Adapt API to new simdrom-core
* Introduce a seed option
* -- name to set a sample-name (overwrites name "Sampled" in vcf file)
* -bGC or -mGC
* het/hom/hemi support
### simdrom-core
* Two bugfixes in spike-in
 * Mutation was not inserted if the variant was the last in a contig
  * Error when two or mutations where inserted at the end of a contig
* big restructuring (using Builders, optionals, etc.)
* Fixing bug in ClinVarFilter (wrong object was used to write down the omim IDs)
* Fixing bugs in VCFSampler and SpikeIn
* Fixing seed option bugs (Collection.shuffle was still random even if a seed was set)
* Limiting possible alleles to max. 2. Something like 2/3/4 should not be possible.
→anymore!
* fix in hardy weinberg
* add possibility to use hom/het/hemi allele counts from exac etc.
* Adding sex to simdrom
 \star user can define the sex of the simulated individual. It can be also be randomly_
→assigned or it can be none (only autosomes)
```

```
* Hardy weinberg sampling works dependend on the sex on X and Y
```

- * Same for ac hom/get/hemi
- * generate genotype not correctly (consider all possible genotype, not only 2, seen \rightarrow at the side when using)
- * adding gnomAD support using the genotype counts (GC) flag
- * htsjdk 2.11.0
- * update ClinVarFilter to support CLNREFSTAT

manual

* creating readthedocs manual!

v0.2

* no changelog available

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Version 3, 29 June 2007

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CHAPTER 2

Indices and tables

- genindex
- modindex
- search