# vcfpy Documentation

Release 0.8.0

**Manuel Holtgrewe** 

October 31, 2016

1	Quick Example	3
2	Features	5
3	Frequently Asked Questions	7

VCFPy is a Python 3 library with good support for both reading and writing VCF files. The documentation is split into three parts (accessible through the navigation on the left):

**Installation & Getting Started** Instructions for the installation of the module and some examples to get you started.

API Documentation This section contains the API documentation for the module

**Project Info** More information on the project, including the changelog, list of contributing authors, and contribution instructions.

# CHAPTER 1

# **Quick Example**

# CHAPTER 2

# **Features**

- Support for reading and writing VCF v4.3
- Interface to INFO and FORMAT fields is based on OrderedDict allows for easier modification than PyVCF (also I find this more pythonic)
- Read (and jump in) and write BGZF files just using vcfpy

6 Chapter 2. Features

# **Frequently Asked Questions**

Why another Python library for VCF? I've been using PyVCF with quite some success in the past. However, the main bottleneck of PyVCF is when you want to modify the per-sample genotype information. There are some issues in the tracker of PyVCF but none of them can really be considered solved. I tried several hours to solve these problems within PyVCF but this never got far or towards a complete rewrite...

For this reason, VCFPy was born and here it is!

- Why Python 3 only? As I'm only using Python 3 code, I see no advantage in carrying around support for legacy Python 2 and maintaining it. At a later point when VCFPy is known to be stable, Python 2 support might be added if someone contributes a pull request.
- What's the state? VCFPy is the result of two full days of development plus some maintenance work later now (right now). I'm using it in several projects but it is not as battle-tested as PyVCF.
- What's the difference to PyVCF? The main difference technical. Instead of uscollections.namedtuple call for storing the annotation, VCFPv uses collections.OrderedDict. This has the advantage that (1) access to optional settings is much more pythonic using .qet (KEY, DEFAULT) instead of qetattr(). Further, (2) adding call annotations (FORMAT) fields is able without any performance penalty where for PyVCF, copy. deepcopy has to be used at some point which is very slow. There has not been any movement in supporting modifying FORMAT fields in PyVCF and here is a library that does this well.
- What's the aim? The aim of the project is to provide simple yet efficient read and write access to VCF files. Eventually, PySAM will probably be a better choice once it has a Python wrapper for the VCF part of htslib. However, as this is still misssing, VCFPy is a good solution for the time being.

# 3.1 Installation

# 3.1.1 Stable release

To install vcfpy, run this command in your terminal:

```
$ pip install vcfpy
```

This is the preferred method to install VCFPy, as it will always install the most recent stable release.

If you don't have pip installed, this Python installation guide can guide you through the process.

# 3.1.2 From sources

The sources for vcfpy can be downloaded from the Github repo.

You can either clone the public repository:

```
$ git clone git://github.com/bihealth/vcfpy
```

Or download the tarball:

```
$ curl -OL https://github.com/bihealth/vcfpy/tarball/master
```

Once you have a copy of the source, you can install it with:

```
$ python setup.py install
```

# 3.2 Getting Started

After installation, you can use VCFPy in your project simply by importing the module.

```
import vcfpy
```

That's all, continue and look at the list of examples.

# 3.3 Examples

This chapter contains several examples for the most important use cases of VCFPy.

# 3.3.1 Reading VCF Files

The following is an example for reading VCF files and writing out a TSV file with the genotype calls of all SNVs. You can find the example Python and VCF file in the sources below the directory examples/vcf\_to\_tsv.

```
#!/usr/bin/env python
# -*- coding: utf-8 -*-
import vcfpy

# Open file, this will read in the header
reader = vcfpy.Reader.from_path('input.vcf')

# Build and print header
header = ['#CHROM', 'POS', 'REF', 'ALT'] + reader.header.samples.names
print('\t'.join(header))

for record in reader:
    if not record.is_snv():
        continue
    line = [record.CHROM, record.POS, record.REF]
    line += [alt.value for alt in record.ALT]
    line += [call.data.get('GT') or './.' for call in record.calls]
    print('\t'.join(map(str, line)))
```

The program call looks as follows.

FORMA

\$ ./vcf_to	_tsv.py							
#CHROM	POS	REF	ALT	BLANK	NA128	378	NA12891	NA12892
chr22	42522392	G	A	0/0	0/1	0/1	0/0	0/0
chr22	42522597	С	T	0/1	0/0	0/0	0/0	0/0
chr22	42522613	G	С	0/1	0/1	0/0	0/1	0/1
chr22	42523003	A	G	0/1	1/1	0/1	0/1	0/1
chr22	42523209	T	С	0/1	1/1	0/1	0/1	0/1
chr22	42523211	T	С	0/0	0/1	0/1	0/0	0/0
chr22	42523409	G	T	0/1	0/1	0/0	0/1	0/1
chr22	42523491	С	T	0/1	0/0	0/0	0/0	0/0
chr22	42523507	A	G	0/1	0/0	0/0	0/0	0/0
chr22	42523805	С	T	0/0	0/0	0/1	0/0	0/0
chr22	42523943	A	G	0/1	1/1	0/1	0/1	0/1
chr22	42524435	T	A	0/1	0/0	0/0	0/0	0/0
[]								

# 3.3.2 Writing VCF Files

The following shows how to add values to the FILTER column to records of an existing VCF file. Adding to existing records is simpler than constructing them from scratch, of course.

The program call looks as follows.

```
##fileformat=VCFv4.3
##contig=<ID=20,length=62435964>
##INFO=<ID=NS, Number=1, Type=Integer, Description="Number of Samples With Data">
##INFO=<ID=DP, Number=1, Type=Integer, Description="Total Depth">
##INFO=<ID=AF, Number=A, Type=Float, Description="Allele Frequency">
##INFO=<ID=AA, Number=1, Type=String, Description="Ancestral Allele">
##INFO=<ID=DB, Number=0, Type=Flag, Description="dbSNP membership, build 129">
##INFO=<ID=H2, Number=0, Type=Flag, Description="HapMap2 membership">
##FILTER=<ID=q10, Description="Quality below 10">
##FILTER=<ID=s50,Description="Less than 50% of samples have data">
##FORMAT=<ID=GT, Number=1, Type=String, Description="Genotype">
##FORMAT=<ID=GQ, Number=1, Type=Integer, Description="Genotype Quality">
##FORMAT=<ID=DP, Number=1, Type=Integer, Description="Read Depth">
##FORMAT=<ID=HQ, Number=2, Type=Integer, Description="Haplotype Quality">
##FILTER=<ID=DP10,Description="total DP < 10">
#CHROM
              POS
                                               ALT
                                                           QUAL
                                                                                       INFO
                          TD
                                    REF
                                                                        FILTER
```

3.3. Examples 9

20	14370	rs6054	1257	G	A	29	PASS	NS=3;DP=	14; AF=0.5; DB
20	17330		T	A	3	q10	NS=3;DP=	11;AF=0.01	7 GT:0
20	1110696	rs60	040355	A	G,T	67	PASS	NS=2	; DP=10; AF=0.3
20	1230237		T		47	PASS	NS=3	;DP=13;AA=	T GT:
20	1234567	mici	rosat1	GTC	G,G	TCT	50 P	ASS;DP10	NS=3;DI

# 3.3.3 Jumping in Tabix-indexed Files

The following shows a small program that extracts a genomic region from the input VCF file and writes it to stdout.

```
#!/usr/bin/env python
# -*- coding: utf-8 -*-
import vcfpy
# Open input, add FILTER header, and open output file
reader = vcfpy.Reader.from_path('input.vcf.gz')
writer = vcfpy.Writer.from_path(
    '/dev/stdout', reader.header, reader.samples)
# Fetch region 20:1,110,694-1,230,237. Note that the coordinates
# in the API call are zero-based and describe half-open intervals.
for record in reader.fetch('20', 1110695, 1230237):
   writer.write_record(record)
```

The program call looks as follows.

```
##fileformat=VCFv4.3
##fileDate=20090805
##source=myImputationProgramV3.1
##reference=file:///seq/references/1000GenomesPilot-NCBI36.fasta
##contig=<ID=1,length=249250621>
##contig=<ID=2,length=243199373>
##contig=<ID=20,length=62435964>
##phasing=partial
##INFO=<ID=NS, Number=1, Type=Integer, Description="Number of Samples With Data">
##INFO=<ID=DP, Number=1, Type=Integer, Description="Total Depth">
##INFO=<ID=AF, Number=A, Type=Float, Description="Allele Frequency">
##INFO=<ID=AA, Number=1, Type=String, Description="Ancestral Allele">
##INFO=<ID=DB, Number=0, Type=Flag, Description="dbSNP membership, build 129">
##INFO=<ID=H2, Number=0, Type=Flaq, Description="HapMap2 membership">
##FILTER=<ID=q10, Description="Quality below 10">
##FILTER=<ID=s50, Description="Less than 50% of samples have data">
##FORMAT=<ID=GT, Number=1, Type=String, Description="Genotype">
##FORMAT=<ID=GQ, Number=1, Type=Integer, Description="Genotype Quality">
##FORMAT=<ID=DP, Number=1, Type=Integer, Description="Read Depth">
##FORMAT=<ID=HQ, Number=2, Type=Integer, Description="Haplotype Quality">
              POS
                         TD
                                   REF
                                                                        FILTER
                                                                                       TNFO
                                                                                                   FORMA
                          rs6040355
                                                                67
                                                                                        NS=2; DP=10; AF=0.3
2.0
          1110696
                                            Α
                                                     G,T
20
          1230237
                                                      47
                                                                             NS=3; DP=13; AA=T
                                   Т
                                                                PASS
```

# 3.4 Best Practice

While not strictly part of the documentation of VCFPy, we include some notes on hints that we consider best practice when building VCF processing applications.

GT:

# 3.4.1 Keep Input Verbatim Where Possible

Try to keep the input verbatim if there is no strong reason for adjusting it. Strong reasons include fixing Type or Number in header lines describing arrays of strings, for example.

Whenever possible, keep the header order intact. VCFPy does this automatically for you (in contrast to PyVCF).

# 3.4.2 Prefer Soft-Filters over Hard-Filters

**Soft**-filters mean annotating your VCF records in the FILTER column whereas **Hard**-filters mean removing records from VCF file. In many situations, it is useful to keep around all VCF records and just annotate why they are to be dropped. Then, in the last step, only the interesting ones are kept.

This makes tracing back easier when and why a record was removed.

# 3.5 Header

### **Contents**

- Header
  - vcfpy.OrderedDict
  - vcfpy.Header
  - vcfpy.HeaderLine
  - vcfpy.header\_without\_lines
  - vcfpy.SimpleHeaderLine
  - vcfpy.AltAlleleHeaderLine
  - vcfpy.MetaHeaderLine
  - vcfpy.PedigreeHeaderLine
  - vcfpy.SampleHeaderLine
  - vcfpy.ContigHeaderLine
  - vcfpy.FilterHeaderLine
  - vcfpy.CompoundHeaderLine
  - vcfpy.InfoHeaderLinevcfpy.FormatHeaderLine
  - vcfpy.FieldInfo
  - vcfpy.SamplesInfos

# 3.5.1 vcfpy.OrderedDict

Convenience export of OrderedDict. When available, the cyordereddict, a Cython-reimplementation of OrderedDict is used for Python before 3.5 (from 3.5, Python ships with a fast, C implementation of OrderedDict).

### class vcfpy.OrderedDict

Dictionary that remembers insertion order

 $clear() \rightarrow None$ . Remove all items from od.

 $copy() \rightarrow a shallow copy of od$ 

**fromkeys**  $(S[, v]) \rightarrow \text{New ordered dictionary with keys from S.}$ 

If not specified, the value defaults to None.

3.5. Header 11

```
items () \rightarrow a set-like object providing a view on D's items
```

**keys** ()  $\rightarrow$  a set-like object providing a view on D's keys

### move\_to\_end()

Move an existing element to the end (or beginning if last==False). Raises KeyError if the element does not exist. When last=True, acts like a fast version of self[key]=self.pop(key).

**pop**  $(k[,d]) \rightarrow v$ , remove specified key and return the corresponding

value. If key is not found, d is returned if given, otherwise KeyError is raised.

**popitem** ()  $\rightarrow$  (k, v), return and remove a (key, value) pair.

Pairs are returned in LIFO order if last is true or FIFO order if false.

**setdefault**  $(k[,d]) \rightarrow \text{od.get}(k,d)$ , also set od[k]=d if k not in od

**update** ([E], \*\*F)  $\rightarrow$  None. Update D from mapping/iterable E and F.

If E present and has a .keys() method, does: for k in E: D[k] = E[k] If E present and lacks .keys() method, does: for (k, v) in E: D[k] = v In either case, this is followed by: for k, v in F.items(): D[k] = v

**values** ()  $\rightarrow$  an object providing a view on D's values

# 3.5.2 vcfpy.Header

class vcfpy.Header (lines=None, samples=None, warning\_helper=<vcfpy.warn\_utils.WarningHelper object>)

Represent header of VCF file

While this class allows mutating records, it should not be changed once it has been assigned to

This class provides function for adding lines to a header and updating the supporting index data structures. There is no explicit API for removing header lines, the best way is to reconstruct a new Header instance with a filtered list of header lines.

### add\_contig\_line (mapping)

Add "contig" header line constructed from the given mapping

**Parameters mapping** — OrderedDict with mapping to add. It is recommended to use OrderedDict over dict as this makes the result reproducible

**Returns** False on conflicting line and True otherwise

### add\_filter\_line(mapping)

Add FILTER header line constructed from the given mapping

Parameters mapping — OrderedDict with mapping to add. It is recommended to use OrderedDict over dict as this makes the result reproducible

Returns False on conflicting line and True otherwise

# add\_format\_line (mapping)

Add FORMAT header line constructed from the given mapping

**Parameters mapping** — OrderedDict with mapping to add. It is recommended to use OrderedDict over dict as this makes the result reproducible

**Returns** False on conflicting line and True otherwise

#### add\_info\_line (mapping)

Add INFO header line constructed from the given mapping

Parameters mapping - OrderedDict with mapping to add. It is recommended to use OrderedDict over dict as this makes the result reproducible

```
Returns False on conflicting line and True otherwise
     add_line (header_line)
          Add header line, updating any necessary support indices
              Returns False on conflicting line and True otherwise
     filter ids()
          Return list of all filter IDs
     format ids()
          Return list of all format IDs
     get_format_field_info(key)
          Return FieldInfo for the given INFO field
     get_info_field_info(key)
          Return FieldInfo for the given INFO field
     get_lines (key)
          Return header lines having the given key as their type
     has_header_line(key, id_)
          Return whether there is a header line with the given ID of the type given by key
              Parameters
                   • key – The VCF header key/line type.
                   • id - The ID value to compare fore
              Returns True if there is a header line starting with ##${key} = in the VCF file having the
                  mapping entry ID set to id_.
     info_ids()
          Return list of all info IDs
     lines = None
          list of :py:HeaderLine objects
     samples = None
          SamplesInfo object
3.5.3 vcfpy.HeaderLine
class vcfpy.HeaderLine (key, value, warning_helper=<vcfpy.warn_utils.WarningHelper object>)
     Base class for VCF header lines
     key = None
          str with key of header line
     serialize()
          Return VCF-serialized version of this header line
     warning_helper = None
```

# 3.5.4 vcfpy.header without lines

Helper for printing warnings

```
vcfpy.header_without_lines (header, remove)
Return Header without lines given in remove
```

3.5. Header 13

remove is an iterable of pairs key/ID with the VCF header key and ID of entry to remove. In the case that a line does not have a mapping entry, you can give the full value to remove.

```
# header is a vcfpy.Header, e.g., as read earlier from file
new_header = vcfpy.without_header_lines(
    header, [('assembly', None), ('FILTER', 'PASS')])
# now, the header lines starting with "##assembly=" and the "PASS"
# filter line will be missing from new_header
```

# 3.5.5 vcfpy.SimpleHeaderLine

Base class for simple header lines, currently contig and filter header lines

Don't use this class directly but rather the sub classes.

Raises vcfpy.exceptions.InvalidHeaderException in the case of missing key "ID"

# mapping = None

collections.OrderedDict with key/value mapping of the attributes

# 3.5.6 vcfpy.AltAlleleHeaderLine

Alternative allele header line

Mostly used for defining symbolic alleles for structural variants and IUPAC ambiguity codes

classmethod from mapping (klass, mapping)

Construct from mapping, not requiring the string value

id = None

name of the alternative allele

# 3.5.7 vcfpy.MetaHeaderLine

Alternative allele header line

Used for defining set of valid values for samples keys

classmethod from\_mapping (klass, mapping)

Construct from mapping, not requiring the string value

id = None

name of the alternative allele

# 3.5.8 vcfpy.PedigreeHeaderLine

Header line for defining a pedigree entry

### classmethod from\_mapping (klass, mapping)

Construct from mapping, not requiring the string value

#### id = None

name of the alternative allele

# 3.5.9 vcfpy.SampleHeaderLine

Header line for defining a SAMPLE entry

#### classmethod from\_mapping (klass, mapping)

Construct from mapping, not requiring the string value

#### id = None

name of the alternative allele

# 3.5.10 vcfpy.ContigHeaderLine

Contig header line

Most importantly, parses the 'length' key into an integer

# classmethod from\_mapping(klass, mapping)

Construct from mapping, not requiring the string value

#### id = None

name of the contig

### length = None

length of the contig, None if missing

# 3.5.11 vcfpy.FilterHeaderLine

FILTER header line

#### description = None

description for the filter, None if missing

### classmethod from\_mapping (klass, mapping)

Construct from mapping, not requiring the string value

id = None

token for the filter

# 3.5.12 vcfpy.CompoundHeaderLine

Base class for compound header lines, currently format and header lines

Compound header lines describe fields that can have more than one entry.

3.5. Header 15

Don't use this class directly but rather the sub classes.

#### mapping = None

OrderedDict with key/value mapping

# 3.5.13 vcfpy.InfoHeaderLine

Header line for INFO fields

Note that the Number field will be parsed into an int if possible. Otherwise, the constants HEADER\_NUMBER\_\* will be used.

### description = None

description, should be given, None if not given

# classmethod from\_mapping (klass, mapping)

Construct from mapping, not requiring the string value

#### id = None

key in the INFO field

#### source = None

source of INFO field, None if not given

### type = None

value type

#### version = None

version of INFO field, None if not given

# 3.5.14 vcfpy.FormatHeaderLine

Header line for FORMAT fields

#### description = None

description, should be given, None if not given

### classmethod from\_mapping (klass, mapping)

Construct from mapping, not requiring the string value

### id = None

key in the INFO field

#### source = None

source of INFO field, None if not given

# type = None

value type

#### version = None

version of INFO field, None if not given

# 3.5.15 vcfpy.FieldInfo

```
class vcfpy.FieldInfo(type_, number, description=None)
    Core information for describing field type and number

description = None
    Description for the header field, optional

number = None
    Number description, either an int or constant

type = None
```

# 3.5.16 vcfpy.SamplesInfos

```
class vcfpy.SamplesInfos (sample_names)
    Helper class for handling and mapping of sample names to numeric indices
    name_to_idx = None
        mapping from sample name to index
    names = None
        list of sample names
```

The type, one of INFO\_TYPES or FORMAT\_TYPES

# 3.6 Input/Output

# **Contents**

- Input/Output
  - vcfpy.Reader
  - vcfpy.Writer

# 3.6.1 vcfpy.Reader

```
class vcfpy.Reader (stream, path=None, tabix_path=None, record_checks=None)
    Class for parsing of files from file-like objects
```

Instead of using the constructor, use the class methods from\_stream() and from\_path().

On construction, the header will be read from the file which can cause problems. After construction, Reader can be used as an iterable of Record.

Raises InvalidHeaderException in the case of problems reading the header

close()

Close underlying stream

fetch (chrom, begin, end)

Jump to the start position of the given chromosomal position and limit iteration to the end position

#### **Parameters**

- **chrom** (str) name of the chromosome to jump to
- **begin** (*int*) 0-based begin position (inclusive)

3.6. Input/Output

• end (int) – 0-based end position (exclusive)

**classmethod from\_path** (*klass*, *path*, *tabix\_path=None*, *record\_checks=None*)

Create new Reader from path

#### **Parameters**

- path the path to load from (converted to str for compatibility with path.py)
- tabix\_path optional string with path to TBI index, automatic inferral from path will be tried on the fly if not given
- record\_checks (list) record checks to perform, can contain 'INFO' and 'FOR-MAT'

classmethod from\_stream(klass, stream, path=None, tabix\_path=None, record\_checks=None)
Create new Reader from file

#### **Parameters**

- stream file-like object to read from
- path optional string with path to store (for display only)
- record\_checks (list) record checks to perform, can contain 'INFO' and 'FOR-MAT'

#### header = None

the Header

#### parser = None

the parser to use

#### path = None

optional str with the path to the stream

#### record checks = None

checks to perform on records, can contain 'FORMAT' and 'INFO'

#### samples = None

the vcfpy.header.SamplesInfos object with the sample name information

#### stream = None

stream (file-like object) to read from

# tabix\_file = None

the pysam. TabixFile used for reading from index bgzip-ed VCF; constructed on the fly

#### tabix\_path = None

optional str with path to tabix file

# 3.6.2 vcfpy.Writer

```
class vcfpy.Writer(stream, header, samples, path=None)
```

Class for writing VCF files to file-like objects

Instead of using the constructor, use the class methods from\_stream() and from\_path().

The writer has to be constructed with a Header object and the full VCF header will be written immediately on construction. This, of course, implies that modifying the header after construction is illegal.

#### close()

Close underlying stream

#### classmethod from\_path (klass, path, header)

Create new Writer from path

#### **Parameters**

- path the path to load from (converted to str for compatibility with path.py)
- header VCF header to use
- samples SamplesInfos to use

classmethod from\_stream(klass, stream, header, path=None, use\_bgzf=None)

Create new Writer from file

Note that for getting bgzf support, you have to pass in a stream opened in binary mode. Further, you either have to provide a path ending in ".gz" or set use\_bgzf=True. Otherwise, you will get the notorious "TypeError: 'str' does not support the buffer interface".

#### **Parameters**

- stream file-like object to write to
- header VCF header to use
- path optional string with path to store (for display only)
- use\_bgzf indicator whether to write bgzf to stream if True, prevent if False, interpret path if None

#### header = None

the :py:class:~vcfpy.header.Header' written out

### path = None

optional str with the path to the stream

# stream = None

stream (file-like object) to read from

#### write record(record)

Write out the given vcfpy.record.Record to this Writer

# 3.7 Exceptions

### **Contents**

- Exceptions
  - vcfpy.VCFPyException
  - vcfpy.InvalidHeaderException
  - $-\ vcfpy. Invalid Record Exception$
  - vcfpy.IncorrectVCFFormat
  - vcfpy.HeaderNotFound

# 3.7.1 vcfpy.VCFPyException

# exception vcfpy.VCFPyException

Base class for module's exception

3.7. Exceptions

# 3.7.2 vcfpy.InvalidHeaderException

**exception** vcfpy.**InvalidHeaderException**Raised in the case of invalid header formatting

# 3.7.3 vcfpy.InvalidRecordException

**exception** vofpy. **InvalidRecordException**Raised in the case of invalid record formatting

# 3.7.4 vcfpy.IncorrectVCFFormat

**exception** vcfpy. **IncorrectVCFFormat**Raised on problems parsing VCF

# 3.7.5 vcfpy.HeaderNotFound

exception vcfpy.HeaderNotFound
Raised when a VCF header could not be found

# 3.8 Records

### **Contents**

- Records
  - Record-Related Constants
  - vcfpy.Record
  - vcfpy.Call
  - vcfpy.AltRecord
  - vcfpy.Substitution
  - vcfpy.SV
  - vcfpy.BreakEnd
  - vcfpy.SingleBreakEnd
  - vcfpy.SymbolicAllele

# 3.8.1 Record-Related Constants

The following constants are also available as vcfpy.CONSTANT.

vcfpy.record. $HOM_REF = 0$ 

Code for homozygous reference

vcfpy.record.HOM\_ALT = 2

Code for homozygous alternative

vcfpy.record.FIVE\_PRIME = '5'

code for five prime orientation BreakEnd

```
vcfpy.record.THREE_PRIME = '3'
    code for three prime orientation BreakEnd
vcfpy.record.FORWARD = '+'
    code for forward orientation
vcfpy.record.REVERSE = '-'
    code for reverse orientation
```

# 3.8.2 vcfpy.Record

# class vcfpy.Record (CHROM, POS, ID, REF, ALT, QUAL, FILTER, INFO, FORMAT, calls)

Represent one record from the VCF file

Record objects are iterators of their calls

#### ALT = None

A list of alternative allele records of type AltRecord

#### CHROM = None

A str with the chromosome name

#### FILTER = None

A list of strings for the FILTER column

#### FORMAT = None

A list of strings for the FORMAT column

#### ID = None

A list of the semicolon-separated values of the ID column

#### INFO = None

An OrderedDict giving the values of the INFO column, flags are mapped to True

#### POS = None

An int with a 1-based begin position

### QUAL = None

The quality value, can be None

#### REF = None

A str with the REF value

#### add\_filter(label)

Add label to FILTER if not set yet, removing PASS entry if present

# add\_format (key, value=None)

Add an entry to format

The record's calls data [key] will be set to value if not yet set and value is not None. If key is already in FORMAT then nothing is done.

### affected\_end

Return affected start position in 0-based coordinates

For SNVs, MNVs, and deletions, the behaviour is based on the start position and the length of the REF. In the case of insertions, the position behind the insert position is returned, yielding a 0-length interval together with affected\_start()

### affected\_start

Return affected start position in 0-based coordinates

3.8. Records 21

For SNVs, MNVs, and deletions, the behaviour is the start position. In the case of insertions, the position behind the insert position is returned, yielding a 0-length interval together with affected\_end()

### begin = None

An int with a 0-based begin position

#### call\_for\_sample = None

A mapping from sample name to entry in self.calls

#### calls = None

A list of genotype Call objects

#### end = None

An int with a 0-based end position

#### is\_snv()

Return True if it is a SNV

# 3.8.3 vcfpy.Call

### class vcfpy.Call (sample, data, site=None)

The information for a genotype callable

By VCF, this should always include the genotype information and can contain an arbitrary number of further annotation, e.g., the coverage at the variant position.

#### called = None

whether or not the variant is fully called

#### data = None

an OrderedDict with the key/value pair information from the call's data

#### gt\_alleles = None

the allele numbers (0, 1, ...) in this calls or None for no-call

#### gt\_bases

Return the actual genotype bases, e.g. if VCF genotype is 0/1, could return ('A', 'T')

# gt\_phase\_char

Return character to use for phasing

#### gt\_type

The type of genotype, returns one of HOM\_REF, HOM\_ALT, and HET.

### is\_filtered(require=None, ignore=None)

Return True for filtered calls

#### **Parameters**

- **ignore** (*iterable*) if set, the filters to ignore, make sure to include 'PASS', when setting, default is ['PASS']
- require (iterable) if set, the filters to require for returning True

### is\_het

Return True for heterozygous calls

# is\_phased

Return boolean indicating whether this call is phased

#### is\_variant

Return True for non-hom-ref calls

#### plodity = None

the number of alleles in this sample's call

### sample = None

the name of the sample for which the call was made

#### site = None

the Record of this Call

# 3.8.4 vcfpy.AltRecord

```
class vcfpy.AltRecord(type_=None)
```

An alternative allele Record

Currently, can be a substitution, an SV placeholder, or breakend

#### serialize()

Return str with representation for VCF file

### type = None

String describing the type of the variant, could be one of SNV, MNV, could be any of teh types described in the ALT header lines, such as DUP, DEL, INS, ...

# 3.8.5 vcfpy.Substitution

```
class vcfpy.Substitution(type_, value)
```

A basic alternative allele record describing a REF->AltRecord substitution

Note that this subsumes MNVs, insertions, and deletions.

#### value = None

The alternative base sequence to use in the substitution

# 3.8.6 vcfpy.SV

vcfpy.SV

# 3.8.7 vcfpy.BreakEnd

A placeholder for a breakend

### mate\_chrom = None

chromosome of the mate breakend

### mate\_orientation = None

orientation breakend's mate

# mate\_pos = None

position of the mate breakend

#### orientation = None

orientation of this breakend

# sequence = None

breakpoint's connecting sequence

3.8. Records 23

#### serialize()

Return string representation for VCF

### within\_main\_assembly = None

bool specifying if the breakend mate is within the assembly (True) or in an ancillary assembly (False)

# 3.8.8 vcfpy.SingleBreakEnd

class vcfpy.SingleBreakEnd(orientation, sequence)

A placeholder for a single breakend

# 3.8.9 vcfpy.SymbolicAllele

#### class vcfpy.SymbolicAllele(value)

A placeholder for a symbolic allele

The allele symbol must be defined in the header using an ALT header before being parsed. Usually, this is used for succinct descriptions of structural variants or IUPAC parameters.

#### value = None

The symbolic value, e.g. 'DUP'

# 3.9 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:

# 3.9.1 Types of Contributions

### **Report Bugs**

Report bugs at https://github.com/bihealth/vcfpy/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. Anything tagged with "bug" and "help wanted" is open to whoever wants to implement it.

#### **Implement Features**

Look through the GitHub issues for features. Anything tagged with "enhancement" and "help wanted" is open to whoever wants to implement it.

### **Write Documentation**

vcfpy 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.

#### **Submit Feedback**

The best way to send feedback is to file an issue at https://github.com/bihealth/vcfpy/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:)

# 3.9.2 Get Started!

Ready to contribute? Here's how to set up vcfpy for local development.

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

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

3. Install your local copy into a virtualenv. Assuming you have virtualenvwrapper installed, this is how you set up your fork for local development:

```
$ mkvirtualenv vcfpy
$ cd vcfpy/
$ python setup.py develop
```

4. 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 changes, check that your changes pass flake8 and the tests, including testing other Python versions with tox:

```
$ flake8 vcfpy tests
$ python setup.py test or py.test
$ tox
```

To get flake8 and tox, just pip install them into your virtualenv.

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

```
$ 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.

3.9. Contributing 25

# 3.9.3 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. Put your new functionality into a function with a docstring, and add the feature to the list in README.rst.
- 3. The pull request should work for Python 3.3, 3.4 and 3.5. Check https://travisci.org/bihealth/vcfpy/pull\_requests and make sure that the tests pass for all supported Python versions.

# 3.9.4 Tips

To run a subset of tests:

\$ py.test tests.test\_vcfpy

# 3.10 Credits

# 3.10.1 Development Lead

• Manuel Holtgrewe <manuel.holtgrewe@bihealth.de>

# 3.10.2 Contributors

None yet. Why not be the first?

# 3.11 History

# 3.11.1 0.8.0 (2016-10-31)

- Adding Header.has\_header\_line for querying existence of header line
- Header.add\_\*\_line return a bool no indicating any conflicts
- Construction of Writer uses samples within header and no extra parameter (breaks API)

# 3.11.2 0.7.0 (2016-09-25)

- Smaller improvements and fixes to documentation
- Adding Codacy coverage and static code analysis results to README
- Various smaller code cleanup triggered by Codacy results
- Adding \_\_eq\_\_, \_\_neq\_\_ and \_\_hash\_\_ to data types (where applicable)

# 3.11.3 0.6.0 (2016-09-25

- · Refining implementation for breakend and symbolic allele class
- Removing record.SV\_CODES
- Refactoring parser module a bit to make the code cleaner
- Fixing small typos and problems in documentation

# 3.11.4 0.5.0 (2016-09-24)

- Deactivating warnings on record parsing by default because of performance
- Adding validation for INFO and FORMAT fields on reading (#8)
- Adding predefined INFO and FORMAT fields to pyvcf.header (#32)

# 3.11.5 0.4.1 (2016-09-22)

• Initially enabling codeclimate

# 3.11.6 0.4.0 (2016-09-22)

- Exporting constants for encoding variant types
- Exporting genotype constants HOM\_REF, HOM\_ALT, HET
- Implementing Call.is\_phased, Call.is\_het, Call.is\_variant, Call.is\_phased, Call.is\_hom\_ref, Call.is\_hom\_alt
- Removing Call.phased (breaks API, next release is 0.4.0)
- Adding tests, fixing bugs for methods of Call

# 3.11.7 0.3.1 (2016-09-21)

• Work around FORMAT/FT being a string; this is done so in the Delly output

# 3.11.8 0.3.0 (2016-09-21)

- Reader and Writer can now be used as context manager (with with)
- Including license in documentation, including Biopython license
- Adding support for writing bgzf files (taken from Biopython)
- Adding support for parsing arrays in header lines
- Removing example-4.1-bnd.vcf example file because v4.1 tumor derival lacks ID field
- Adding AltAlleleHeaderLine, MetaHeaderLine, PedigreeHeaderLine, and SampleHeaderLine
- Renaming SimpleHeaderFile to SimpleHeaderLine
- Warn on missing FILTER entries on parsing

3.11. History 27

- Reordered parameters in from\_stream and from\_file (#18)
- Renamed from\_file to from\_stream (#18)
- Renamed Reader.jump\_to to Reader.fetch
- Adding header\_without\_lines function
- Generally extending API to make it esier to use
- Upgrading dependencies, enabling pyup-bot
- · Greatly extending documentation

# 3.11.9 0.2.1 (2016-09-19)

· First release on PyPI

# 3.12 License

# 3.12.1 VCFPy License

You can find the License of VCFPy below.

MIT License

Copyright (c) 2016, Berlin Institute of Health

Permission is hereby granted, free of charge, to any person obtaining a copy of this software and associated documentation files (the "Software"), to deal in the Software without restriction, including without limitation the rights to use, copy, modify, merge, publish, distribute, sublicense, and/or sell copies of the Software, and to permit persons to whom the Software is furnished to do so, subject to the following conditions:

The above copyright notice and this permission notice shall be included in all copies or substantial portions of the Software.

THE SOFTWARE IS PROVIDED "AS IS", WITHOUT WARRANTY OF ANY KIND, EXPRESS OR IMPLIED, INCLUDING BUT NOT LIMITED TO THE WARRANTIES OF MERCHANTABILITY, FITNESS FOR A PARTICULAR PURPOSE AND NONINFRINGEMENT. IN NO EVENT SHALL THE AUTHORS OR COPYRIGHT HOLDERS BE LIABLE FOR ANY CLAIM, DAMAGES OR OTHER LIABILITY, WHETHER IN AN ACTION OF CONTRACT, TORT OR OTHERWISE, ARISING FROM, OUT OF OR IN CONNECTION WITH THE SOFTWARE OR THE USE OR OTHER DEALINGS IN THE SOFTWARE.

# 3.12.2 Biopython License Agreement

The bgzf writing code is taken from the Biopython project. You can find a copy of the license below.

Biopython License Agreement

Permission to use, copy, modify, and distribute this software and its documentation with or without modifications and for any purpose and without fee is hereby granted, provided that any copyright notices appear in all copies and that both those copyright notices and this

permission notice appear in supporting documentation, and that the names of the contributors or copyright holders not be used in advertising or publicity pertaining to distribution of the software without specific prior permission.

THE CONTRIBUTORS AND COPYRIGHT HOLDERS OF THIS SOFTWARE DISCLAIM ALL WARRANTIES WITH REGARD TO THIS SOFTWARE, INCLUDING ALL IMPLIED WARRANTIES OF MERCHANTABILITY AND FITNESS, IN NO EVENT SHALL THE CONTRIBUTORS OR COPYRIGHT HOLDERS BE LIABLE FOR ANY SPECIAL, INDIRECT OR CONSEQUENTIAL DAMAGES OR ANY DAMAGES WHATSOEVER RESULTING FROM LOSS OF USE, DATA OR PROFITS, WHETHER IN AN ACTION OF CONTRACT, NEGLIGENCE OR OTHER TORTIOUS ACTION, ARISING OUT OF OR IN CONNECTION WITH THE USE OR PERFORMANCE OF THIS SOFTWARE.

3.12. License 29

A	F
add_contig_line() (vcfpy.Header method), 12 add_filter() (vcfpy.Record method), 21 add_filter_line() (vcfpy.Header method), 12 add_format() (vcfpy.Record method), 21 add_format_line() (vcfpy.Header method), 12 add_info_line() (vcfpy.Header method), 12 add_line() (vcfpy.Header method), 13 affected_end (vcfpy.Record attribute), 21 affected_start (vcfpy.Record attribute), 21 ALT (vcfpy.Record attribute), 21 AltAlleleHeaderLine (class in vcfpy), 14 AltRecord (class in vcfpy), 23	fetch() (vcfpy.Reader method), 17 FieldInfo (class in vcfpy), 17 FILTER (vcfpy.Record attribute), 21 filter_ids() (vcfpy.Header method), 13 FilterHeaderLine (class in vcfpy), 15 FORMAT (vcfpy.Record attribute), 21 format_ids() (vcfpy.Header method), 13 FormatHeaderLine (class in vcfpy), 16 from_mapping() (vcfpy.AltAlleleHeaderLine class method), 14 from_mapping() (vcfpy.ContigHeaderLine class method), 15
В	from_mapping() (vcfpy.FilterHeaderLine class method),  15
begin (vcfpy.Record attribute), 22 BreakEnd (class in vcfpy), 23	from_mapping() (vcfpy.FormatHeaderLine class method), 16  from_mapping() (vcfpy.InfeHeaderLine class method)
Call (class in vcfpy), 22 call_for_sample (vcfpy.Record attribute), 22 called (vcfpy.Call attribute), 22 calls (vcfpy.Record attribute), 22 calls (vcfpy.Record attribute), 22 CHROM (vcfpy.Record attribute), 21 clear() (vcfpy.OrderedDict method), 11 close() (vcfpy.Reader method), 17 close() (vcfpy.Writer method), 18 CompoundHeaderLine (class in vcfpy), 15 ContigHeaderLine (class in vcfpy), 15 copy() (vcfpy.OrderedDict method), 11	from_mapping() (vcfpy.InfoHeaderLine class method),  16  from_mapping() (vcfpy.MetaHeaderLine class method),  14  from_mapping() (vcfpy.PedigreeHeaderLine class method), 14  from_mapping() (vcfpy.SampleHeaderLine class method), 15  from_path() (vcfpy.Reader class method), 18  from_path() (vcfpy.Writer class method), 18  from_stream() (vcfpy.Reader class method), 18  from_stream() (vcfpy.Writer class method), 19  fromkeys() (vcfpy.OrderedDict method), 11
D data (vcfpy.Call attribute), 22 description (vcfpy.FieldInfo attribute), 17 description (vcfpy.FilterHeaderLine attribute), 15 description (vcfpy.FormatHeaderLine attribute), 16 description (vcfpy.InfoHeaderLine attribute), 16	get_format_field_info() (vcfpy.Header method), 13 get_info_field_info() (vcfpy.Header method), 13 get_lines() (vcfpy.Header method), 13 gt_alleles (vcfpy.Call attribute), 22 gt_bases (vcfpy.Call attribute), 22 gt_phase_char (vcfpy.Call attribute), 22 gt_type (vcfpy.Call attribute), 22
end (vcfny.Record attribute). 22	

Н	0
has_header_line() (vcfpy.Header method), 13	OrderedDict (class in vcfpy), 11
Header (class in vcfpy), 12	orientation (vcfpy.BreakEnd attribute), 23
header (vcfpy.Reader attribute), 18	
header (vcfpy.Writer attribute), 19	P
header_without_lines() (in module vcfpy), 13	parser (vcfpy.Reader attribute), 18
HeaderLine (class in vcfpy), 13	path (vcfpy.Reader attribute), 18
HeaderNotFound, 20	path (vcfpy.Writer attribute), 19
1	PedigreeHeaderLine (class in vcfpy), 14
id (vcfpy.AltAlleleHeaderLine attribute), 14	plodity (vcfpy.Call attribute), 22
id (vcfpy.ContigHeaderLine attribute), 15	pop() (vcfpy.OrderedDict method), 12 popitem() (vcfpy.OrderedDict method), 12
id (vcfpy.FilterHeaderLine attribute), 15	POS (vcfpy.Record attribute), 21
id (vcfpy.FormatHeaderLine attribute), 16	Tob (verpy.needra antibate), 21
id (vcfpy.InfoHeaderLine attribute), 16	Q
id (vcfpy.MetaHeaderLine attribute), 14	QUAL (vcfpy.Record attribute), 21
id (vcfpy.PedigreeHeaderLine attribute), 15	
ID (vcfpy.Record attribute), 21	R
id (vcfpy.SampleHeaderLine attribute), 15 IncorrectVCFFormat, 20	Reader (class in vcfpy), 17
INFO (vcfpy.Record attribute), 21	Record (class in vcfpy), 21
info_ids() (vcfpy.Header method), 13	record_checks (vcfpy.Reader attribute), 18
InfoHeaderLine (class in vcfpy), 16	REF (vcfpy.Record attribute), 21
InvalidHeaderException, 20	S
InvalidRecordException, 20	
is_filtered() (vcfpy.Call method), 22	sample (vcfpy.Call attribute), 23 SampleHeaderLine (class in vcfpy), 15
is_het (vcfpy.Call attribute), 22	samples (vcfpy.Header attribute), 13
is_phased (vcfpy.Call attribute), 22 is_snv() (vcfpy.Record method), 22	samples (vcfpy.Reader attribute), 18
is_variant (vcfpy.Call attribute), 22	SamplesInfos (class in vcfpy), 17
items() (vcfpy.OrderedDict method), 11	sequence (vcfpy.BreakEnd attribute), 23
	serialize() (vcfpy.AltRecord method), 23
K	serialize() (vcfpy.BreakEnd method), 24
key (vcfpy.HeaderLine attribute), 13	serialize() (vcfpy.HeaderLine method), 13
keys() (vcfpy.OrderedDict method), 12	setdefault() (vcfpy.OrderedDict method), 12 SimpleHeaderLine (class in vcfpy), 14
1	SingleBreakEnd (class in verpy), 24
L	site (vcfpy.Call attribute), 23
length (vcfpy.ContigHeaderLine attribute), 15	source (vcfpy.FormatHeaderLine attribute), 16
lines (vcfpy.Header attribute), 13	source (vcfpy.InfoHeaderLine attribute), 16
M	stream (vcfpy.Reader attribute), 18
mapping (vcfpy.CompoundHeaderLine attribute), 16	stream (vcfpy.Writer attribute), 19
mapping (verpy. Compounds reduct Eine attribute), 14	Substitution (class in vcfpy), 23 SV (in module vcfpy), 23
mate_chrom (vcfpy.BreakEnd attribute), 23	Symbolic Allele (class in vcfpy), 24
mate_orientation (vcfpy.BreakEnd attribute), 23	Symbolic mele (class in verpy), 21
mate_pos (vcfpy.BreakEnd attribute), 23	Τ
MetaHeaderLine (class in vcfpy), 14	tabix_file (vcfpy.Reader attribute), 18
move_to_end() (vcfpy.OrderedDict method), 12	tabix_path (vcfpy.Reader attribute), 18
N	type (vcfpy.AltRecord attribute), 23
	type (vcfpy.FieldInfo attribute), 17
name_to_idx (vcfpy.SamplesInfos attribute), 17 names (vcfpy.SamplesInfos attribute), 17	type (vcfpy.FormatHeaderLine attribute), 16
number (vcfpy.FieldInfo attribute), 17	type (vcfpy.InfoHeaderLine attribute), 16

32 Index

# U

update() (vcfpy.OrderedDict method), 12

# V

value (vcfpy.Substitution attribute), 23 value (vcfpy.SymbolicAllele attribute), 24 values() (vcfpy.OrderedDict method), 12 VCFPyException, 19 version (vcfpy.FormatHeaderLine attribute), 16 version (vcfpy.InfoHeaderLine attribute), 16

# W

warning\_helper (vcfpy.HeaderLine attribute), 13 within\_main\_assembly (vcfpy.BreakEnd attribute), 24 write\_record() (vcfpy.Writer method), 19 Writer (class in vcfpy), 18

Index 33