vcfpy Documentation

Release 0.11.2+0.g697768d.dirty

Manuel Holtgrewe

Apr 16, 2018

Installation Getting Started

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

```
#!/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')
reader.header.add_filter_line(vcfpy.OrderedDict([
    ('ID', 'DP10'), ('Description', 'total DP < 10')]))
writer = vcfpy.Writer.from_path('/dev/stdout', reader.header)
# Add "DP10" filter to records having less than 10 reads
for record in reader:
    ad = sum(c.data.get('DP', 0) for c in record.calls)
    if ad < 10:
        record.add_filter('DP10')
    writer.write_record(record)</pre>
```

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 ${\tt vcfpy}$

CHAPTER 3

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 is technical. Instead of using collections. namedtuple for storing the call annotation, VCFPy uses collections.OrderedDict. This has the advantage that (1) access to optional settings is much more pythonic using .get(KEY, DEFAULT) instead of getattr(). 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.

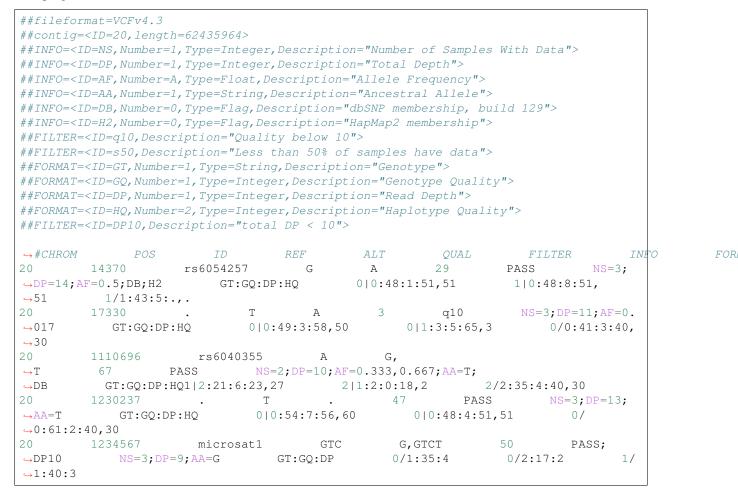
\$./vcf_tc	_tsv.py								
→ #CHROM	POS		REF		ALT	BLANK	NA1	2878	NA12891
chr22	42522392		G		A	0/0	0/1	0/1	0/
⇔0	0/0	0/0		0/0					
chr22	42522597		С		Т	0/1	0/0	0/0	0/
∽0	0/0	0/0		0/0					
chr22	42522613		G		С	0/1	0/1	0/0	0/
⇔1	0/1	0/1		0/1					
chr22	42523003		A		G	0/1	1/1	0/1	0/
<u>⇔</u> 1	0/1	0/1		0/1					
chr22	42523209		Т		С	0/1	1/1	0/1	0/
	0/1								
	42523211					0/0	0/1	0/1	0/
⇔ 0	0/0	0/0		0/0					
chr22	42523409		-		Т	0/1	0/1	0/0	0/
	0/1			0/1					
	42523491				Т	0/1	0/0	0/0	0/
	0/0								
	42523507					0/1	0/0	0/0	0/
↔ 0	0/0			0/0					
chr22	42523805					0/0	0/0	0/1	0/
	0/0								
	42523943				G	0/1	1/1	0/1	0/
	0/1			0/1					
chr22	42524435				A	0/1	0/0	0/0	0/
∽ 0	0/0	0/1		0/1					
[]									

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.

```
#!/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')
reader.header.add_filter_line(vcfpy.OrderedDict([
    ('ID', 'DP10'), ('Description', 'total DP < 10')]))
writer = vcfpy.Writer.from_path('/dev/stdout', reader.header)
# Add "DP10" filter to records having less than 10 reads
for record in reader:
    ad = sum(c.data.get('DP', 0) for c in record.calls)
    if ad < 10:
        record.add_filter('DP10')
    writer.write_record(record)</pre>
```

The program call looks as follows.



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)
# 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=Flag,Description="HapMap2 membership">
##FILTER=<ID=g10, 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
                                                                                     TNFO
\hookrightarrow #CHROM
                          ID
                                    REF
                                               AT_{t}T
                                                           OUAL
                                                                      FILTER
20
        1110696
                       rs6040355
                                         А
                                                  G,
                    PASS NS=2; DP=10; AF=0.333, 0.667; AA=T;
⇔Τ
         67
⇔DB
          GT:GQ:DP:HQ1|2:21:6:23,27
                                         2|1:2:0:18,2
                                                                2/2:35:4:40,30
20
         1230237 .
                                Т
                                             47
                                                             PASS
                                                                       NS=3;DP=13;
                               0|0:54:7:56,60
 \rightarrow AA = T 
             GT:GQ:DP:HQ
                                                      0|0:48:4:51,51
                                                                             0/
↔0:61:2:40,30
```

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.

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

FOR



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

 $\operatorname{{\tt clear}}$ () \rightarrow None. Remove all items from od.

 $\operatorname{\textbf{copy}}$ () \rightarrow a shallow copy of od

fromkeys $(S[, v]) \rightarrow$ New ordered dictionary with keys from S. If not specified, the value defaults to None.

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.
```

 $\begin{array}{l} \textbf{popitem}\,(\,)\,\rightarrow(k,\,v),\,return\,\,and\,\,remove\,\,a\,\,(key,\,value)\,\,pair.\\ Pairs\,\,are\,\,returned\,\,in\,\,LIFO\,\,order\,\,if\,\,last\,\,is\,\,true\,\,or\,\,FIFO\,\,order\,\,if\,\,false. \end{array}$

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

3.5.2 vcfpy.Header

```
class vcfpy.Header(lines=None, samples=None)
```

Represent header of VCF file

While this class allows mutating records, it should not be changed once it has been assigned to a writer. Use **:py:method: `~Header.copy'** to create a copy that can be modified without problems.

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

copy()

Return a copy of this header

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)
```

Base class for VCF header lines

copy () Return a copy

```
key = None
```

str with key of header line

```
serialize()
```

Return VCF-serialized version of this header line

3.5.4 vcfpy.header_without_lines

```
vcfpy.header_without_lines(header, remove)
```

Return Header without lines given in remove

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.

3.5.5 vcfpy.SimpleHeaderLine

```
class vcfpy.SimpleHeaderLine(key, value, mapping)
```

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"

```
copy()
```

Return a copy

```
mapping = None
```

collections.OrderedDict with key/value mapping of the attributes

3.5.6 vcfpy.AltAlleleHeaderLine

```
class vcfpy.AltAlleleHeaderLine(key, value, mapping)
```

Alternative allele header line

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

```
classmethod from_mapping(mapping)
```

Construct from mapping, not requiring the string value

id = None
 name of the alternative allele

3.5.7 vcfpy.MetaHeaderLine

```
class vcfpy.MetaHeaderLine (key, value, mapping)
Alternative allele header line
```

Used for defining set of valid values for samples keys

```
classmethod from_mapping (mapping)
Construct from mapping, not requiring the string value
```

id = None

name of the alternative allele

3.5.8 vcfpy.PedigreeHeaderLine

```
class wcfpy.PedigreeHeaderLine (key, value, mapping)
Header line for defining a pedigree entry
```

```
classmethod from_mapping (mapping)
Construct from mapping, not requiring the string value
```

id = None

name of the alternative allele

3.5.9 vcfpy.SampleHeaderLine

```
class vcfpy.SampleHeaderLine (key, value, mapping)
Header line for defining a SAMPLE entry
```

classmethod from_mapping (*mapping*) Construct from mapping, not requiring the string value

id = None

name of the alternative allele

3.5.10 vcfpy.ContigHeaderLine

```
class vcfpy.ContigHeaderLine(key, value, mapping)
        Contig header line
```

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

```
classmethod from_mapping (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

```
class vcfpy.FilterHeaderLine (key, value, mapping)
FILTER header line
```

- description = None description for the filter, None if missing
- **classmethod from_mapping** (*mapping*) Construct from mapping, not requiring the string value
- id = None token for the filter

3.5.12 vcfpy.CompoundHeaderLine

```
class vcfpy.CompoundHeaderLine(key, value, mapping)
```

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

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

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

copy () Return a copy

```
mapping = None
```

OrderedDict with key/value mapping

3.5.13 vcfpy.InfoHeaderLine

```
class vcfpy.InfoHeaderLine (key, value, mapping)
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(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

```
class vcfpy.FormatHeaderLine(key, value, mapping)
    Header line for FORMAT fields
    description = None
```

description, should be given, None if not given

```
classmethod from_mapping (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, id_=None)
Core information for describing field type and number
```

description = None Description for the header field, optional

id = None

The id of the field, optional.

number = None

Number description, either an int or constant

```
type = None
```

The type, one of INFO_TYPES or FORMAT_TYPES

3.5.16 vcfpy.SamplesInfos

class vcfpy.**SamplesInfos** (*sample_names*, *parsed_samples=None*) Helper class for handling the samples in VCF files

The purpose of this class is to decouple the sample name list somewhat from *Header*. This encapsulates subsetting samples for which the genotype should be parsed and reordering samples into output files.

Note that when subsetting is used and the records are to be written out again then the FORMAT field must not be touched.

copy()

Return a copy of the object

```
is_parsed(name)
```

Return whether the sample name is parsed

```
name_to_idx = None
```

mapping from sample name to index

names = None

list of sample that are read from/written to the VCF file at hand in the given order

parsed_samples = None

set with the samples for which the genotype call fields should be read; can be used for partial parsing (speedup) and defaults to the full list of samples, None if all are parsed

3.6 Input/Output

Contents Input/Output vcfpy.Reader vcfpy.Writer

3.6.1 vcfpy.Reader

tabix_path=None,

record_checks=None,

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

Note: It is important to note that the header member is used during the parsing of the file. If you need a modified version then create a copy, e.g., using :py:method:'~vcfpy.header.Header.copy'.

Note: If you use the parsed_samples feature and you write out records then you must not change the FORMAT of the record.

close()

Close underlying stream

fetch (chrom_or_region, begin=None, end=None)

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

Parameters

- chrom_or_region (str) name of the chromosome to jump to if begin and end are given and a samtools region string otherwise (e.g. "chr1:123,456-123,900").
- **begin** (*int*) 0-based begin position (inclusive)
- **end** (*int*) 0-based end position (exclusive)

Note: If you use the parsed_samples feature and you write out records then you must not change the FORMAT of the record.

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'

Create new Reader from file

Note: If you use the parsed_samples feature and you write out records then you must not change the FORMAT of the record.

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'
- **parsed_samples** (*list*) list of str values with names of samples to parse call information for (for speedup); leave to None for ignoring

header = None

the Header

classmethod from_path (path, tabix_path=None, record_checks=None, parsed_samples=None)
Create new Reader from path

```
parsed_samples = None
```

if set, list of samples to parse for

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'

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, 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.

$\verb+close()$

Close underlying stream

classmethod from_path (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, lines and samples are deep-copied

classmethod from_stream(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, lines and samples are deep-copied
- **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' to write out, will be deep-copied into the Writer on initialization

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.InvalidRecordException
 - vcfpy.IncorrectVCFFormat
 - vcfpy.HeaderNotFound

3.7.1 vcfpy.VCFPyException

exception vcfpy.VCFPyException Base class for module's exception

3.7.2 vcfpy.InvalidHeaderException

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

3.7.3 vcfpy.InvalidRecordException

```
exception vcfpy.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



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.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

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

$\texttt{is_snv}()$

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

class vcfpy.**BreakEnd** (*mate_chrom*, *mate_pos*, *orientation*, *mate_orientation*, *sequence*, *within_main_assembly*) A placeholder for a breakend

A placeholder for a breakend

mate_chrom = None
 chromosome of the mate breakend

mate_pos = None
 position of the mate breakend

orientation = None orientation of this breakend

sequence = None
 breakpoint's connecting sequence

```
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.

3.9.3 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://travis-ci.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 v0.11.2 (2018-04-16)

• Removing pip module from setup.py which is not recommended anyway.

3.11.2 v0.11.1 (2018-03-06)

• Working around problem in HTSJDK output with incomplete FORMAT fields (#127). Writing out . instead of keeping trailing empty records empty.

3.11.3 v0.11.0 (2017-11-22)

- The field FORMAT/FT is now expected to be a semicolon-separated string. Internally, we will handle it as a list.
- Switching from warning helper utility code to Python warnings module.
- Return str in case of problems with parsing value.

3.11.4 v0.10.0 (2017-02-27)

- Extending API to allow for reading subsets of records. (Writing for sample subsets or reordered samples is possible through using the appropriate names list in the SamplesInfos for the Writer).
- Deep-copying header lines and samples infos on Writer construction
- Using samples attribute from Header in Reader and Writer instead of passing explicitely

3.11.5 0.9.0 (2017-02-26)

- Restructuring of requirements.txt files
- Fixing parsing of no-call GT fields

3.11.6 0.8.1 (2017-02-08)

- PEP8 style adjustments
- Using versioneer for versioning
- Using requirements *.txt files now from setup.py
- Fixing dependency on cyordereddict to be for Python <3.6 instead of <3.5
- Jumping by samtools coordinate string now also allowed

3.11.7 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)

3.11.8 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)

3.11.9 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

3.11.10 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)

3.11.11 0.4.1 (2016-09-22)

• Initially enabling codeclimate

3.11.12 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

3.11.13 0.3.1 (2016-09-21)

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

3.11.14 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
- 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

3.11.15 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.

Index

A

add_contig_line() (vcfpy.Header method), 13 add_filter() (vcfpy.Record method), 23 add_filter_line() (vcfpy.Header method), 13 add_format() (vcfpy.Record method), 23 add_format_line() (vcfpy.Header method), 13 add_info_line() (vcfpy.Header method), 13 add_line() (vcfpy.Header method), 13 affected_end (vcfpy.Record attribute), 23 affected_start (vcfpy.Record attribute), 23 ALT (vcfpy.Record attribute), 22 AltAlleleHeaderLine (class in vcfpy), 15 AltRecord (class in vcfpy), 24

В

begin (vcfpy.Record attribute), 23 BreakEnd (class in vcfpy), 25

С

Call (class in vcfpy), 23 call for sample (vcfpy.Record attribute), 23 called (vcfpy.Call attribute), 24 calls (vcfpy.Record attribute), 23 CHROM (vcfpy.Record attribute), 22 clear() (vcfpy.OrderedDict method), 12 close() (vcfpy.Reader method), 19 close() (vcfpy.Writer method), 20 CompoundHeaderLine (class in vcfpy), 16 ContigHeaderLine (class in vcfpy), 16 copy() (vcfpy.CompoundHeaderLine method), 16 copy() (vcfpy.Header method), 13 copy() (vcfpy.HeaderLine method), 14 copy() (vcfpy.OrderedDict method), 12 copy() (vcfpy.SamplesInfos method), 18 copy() (vcfpy.SimpleHeaderLine method), 15

D

data (vcfpy.Call attribute), 24 description (vcfpy.FieldInfo attribute), 17 description (vcfpy.FilterHeaderLine attribute), 16 description (vcfpy.FormatHeaderLine attribute), 17 description (vcfpy.InfoHeaderLine attribute), 17

Ε

end (vcfpy.Record attribute), 23

F

fetch() (vcfpy.Reader method), 19 FieldInfo (class in vcfpy), 17 FILTER (vcfpy.Record attribute), 22 filter ids() (vcfpy.Header method), 13 FilterHeaderLine (class in vcfpy), 16 FORMAT (vcfpy.Record attribute), 23 format ids() (vcfpy.Header method), 13 FormatHeaderLine (class in vcfpy), 17 from_mapping() (vcfpy.AltAlleleHeaderLine class method), 15 from_mapping() (vcfpy.ContigHeaderLine class method), 16 from_mapping() (vcfpy.FilterHeaderLine class method), 16 from_mapping() (vcfpy.FormatHeaderLine class method), 17 from_mapping() (vcfpy.InfoHeaderLine class method), 17 from_mapping() (vcfpy.MetaHeaderLine class method), 15 from mapping() (vcfpy.PedigreeHeaderLine class method), 15 from mapping() (vcfpy.SampleHeaderLine class method), 15 from_path() (vcfpy.Reader class method), 19 from_path() (vcfpy.Writer class method), 20 from_stream() (vcfpy.Reader class method), 19 from_stream() (vcfpy.Writer class method), 20 fromkeys() (vcfpy.OrderedDict method), 12

G

get_format_field_info() (vcfpy.Header method), 13

get_info_field_info() (vcfpy.Header method), 13 get_lines() (vcfpy.Header method), 14 gt_alleles (vcfpy.Call attribute), 24 gt_bases (vcfpy.Call attribute), 24 gt_phase_char (vcfpy.Call attribute), 24 gt_type (vcfpy.Call attribute), 24

Η

has_header_line() (vcfpy.Header method), 14 Header (class in vcfpy), 13 header (vcfpy.Reader attribute), 19 header (vcfpy.Writer attribute), 20 header_without_lines() (in module vcfpy), 14 HeaderLine (class in vcfpy), 14 HeaderNotFound, 21

I

id (vcfpy.AltAlleleHeaderLine attribute), 15 id (vcfpy.ContigHeaderLine attribute), 16 id (vcfpy.FieldInfo attribute), 17 id (vcfpy.FilterHeaderLine attribute), 16 id (vcfpy.FormatHeaderLine attribute), 17 id (vcfpy.InfoHeaderLine attribute), 17 id (vcfpy.MetaHeaderLine attribute), 15 id (vcfpy.PedigreeHeaderLine attribute), 15 ID (vcfpy.Record attribute), 23 id (vcfpy.SampleHeaderLine attribute), 16 IncorrectVCFFormat, 21 INFO (vcfpy.Record attribute), 23 info_ids() (vcfpy.Header method), 14 InfoHeaderLine (class in vcfpy), 16 InvalidHeaderException, 21 InvalidRecordException, 21 is_filtered() (vcfpy.Call method), 24 is_het (vcfpy.Call attribute), 24 is_parsed() (vcfpy.SamplesInfos method), 18 is_phased (vcfpy.Call attribute), 24 is snv() (vcfpy.Record method), 23 is variant (vcfpy.Call attribute), 24

K

key (vcfpy.HeaderLine attribute), 14

L

length (vcfpy.ContigHeaderLine attribute), 16 lines (vcfpy.Header attribute), 14

Μ

mapping (vcfpy.CompoundHeaderLine attribute), 16 mapping (vcfpy.SimpleHeaderLine attribute), 15 mate_chrom (vcfpy.BreakEnd attribute), 25 mate_orientation (vcfpy.BreakEnd attribute), 25 mate_pos (vcfpy.BreakEnd attribute), 25 MetaHeaderLine (class in vcfpy), 15 move_to_end() (vcfpy.OrderedDict method), 12

Ν

name_to_idx (vcfpy.SamplesInfos attribute), 18 names (vcfpy.SamplesInfos attribute), 18 number (vcfpy.FieldInfo attribute), 17

0

OrderedDict (class in vcfpy), 12 orientation (vcfpy.BreakEnd attribute), 25

Ρ

parsed_samples (vcfpy.Reader attribute), 19 parsed_samples (vcfpy.SamplesInfos attribute), 18 parser (vcfpy.Reader attribute), 20 path (vcfpy.Reader attribute), 20 path (vcfpy.Writer attribute), 20 PedigreeHeaderLine (class in vcfpy), 15 plodity (vcfpy.Call attribute), 24 pop() (vcfpy.OrderedDict method), 12 popitem() (vcfpy.OrderedDict method), 12 POS (vcfpy.Record attribute), 23

Q

QUAL (vcfpy.Record attribute), 23

R

Reader (class in vcfpy), 18 Record (class in vcfpy), 22 record_checks (vcfpy.Reader attribute), 20 REF (vcfpy.Record attribute), 23

S

sample (vcfpy.Call attribute), 24 SampleHeaderLine (class in vcfpy), 15 samples (vcfpy.Header attribute), 14 SamplesInfos (class in vcfpy), 18 sequence (vcfpy.BreakEnd attribute), 25 serialize() (vcfpy.AltRecord method), 24 serialize() (vcfpy.BreakEnd method), 25 serialize() (vcfpy.HeaderLine method), 14 setdefault() (vcfpy.OrderedDict method), 12 SimpleHeaderLine (class in vcfpy), 15 SingleBreakEnd (class in vcfpy), 25 site (vcfpy.Call attribute), 24 source (vcfpy.FormatHeaderLine attribute), 17 source (vcfpy.InfoHeaderLine attribute), 17 stream (vcfpy.Reader attribute), 20 stream (vcfpy.Writer attribute), 21 Substitution (class in vcfpy), 25 SV (in module vcfpy), 25 SymbolicAllele (class in vcfpy), 25

Т

tabix_file (vcfpy.Reader attribute), 20 tabix_path (vcfpy.Reader attribute), 20 type (vcfpy.AltRecord attribute), 24 type (vcfpy.FieldInfo attribute), 17 type (vcfpy.FormatHeaderLine attribute), 17 type (vcfpy.InfoHeaderLine attribute), 17

V

value (vcfpy.Substitution attribute), 25 value (vcfpy.SymbolicAllele attribute), 25 VCFPyException, 21 version (vcfpy.FormatHeaderLine attribute), 17 version (vcfpy.InfoHeaderLine attribute), 17

W

within_main_assembly (vcfpy.BreakEnd attribute), 25 write_record() (vcfpy.Writer method), 21 Writer (class in vcfpy), 20