

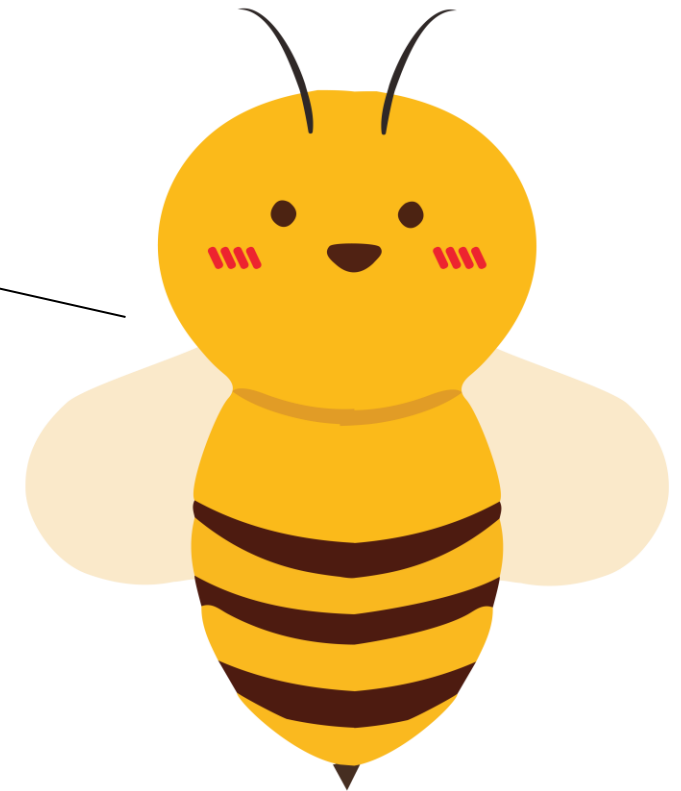


If you are a Linux or a MacOS user, [continue to slide 3](#).

If you are running Windows - bring some coffee,
we have some prerequisites to prepare.

This will take several minutes –

but with perseverance, [glory awaits at slide 4](#)!





Installation For Linux/macOS users

Via terminal, install needed Python 3 packages using the command line:

```
pip3 install pandas dash lxml html5lib
```

Proceed to install samtools:

Linux command > `sudo apt-get install samtools`

MacOS command > `brew install samtools`

That's it, [let's jump to slide 14.](#)



Installation For Windows users

You need Python 3 to run BAMdelbee.

If you already have it installed, [jump slide 7.](#)

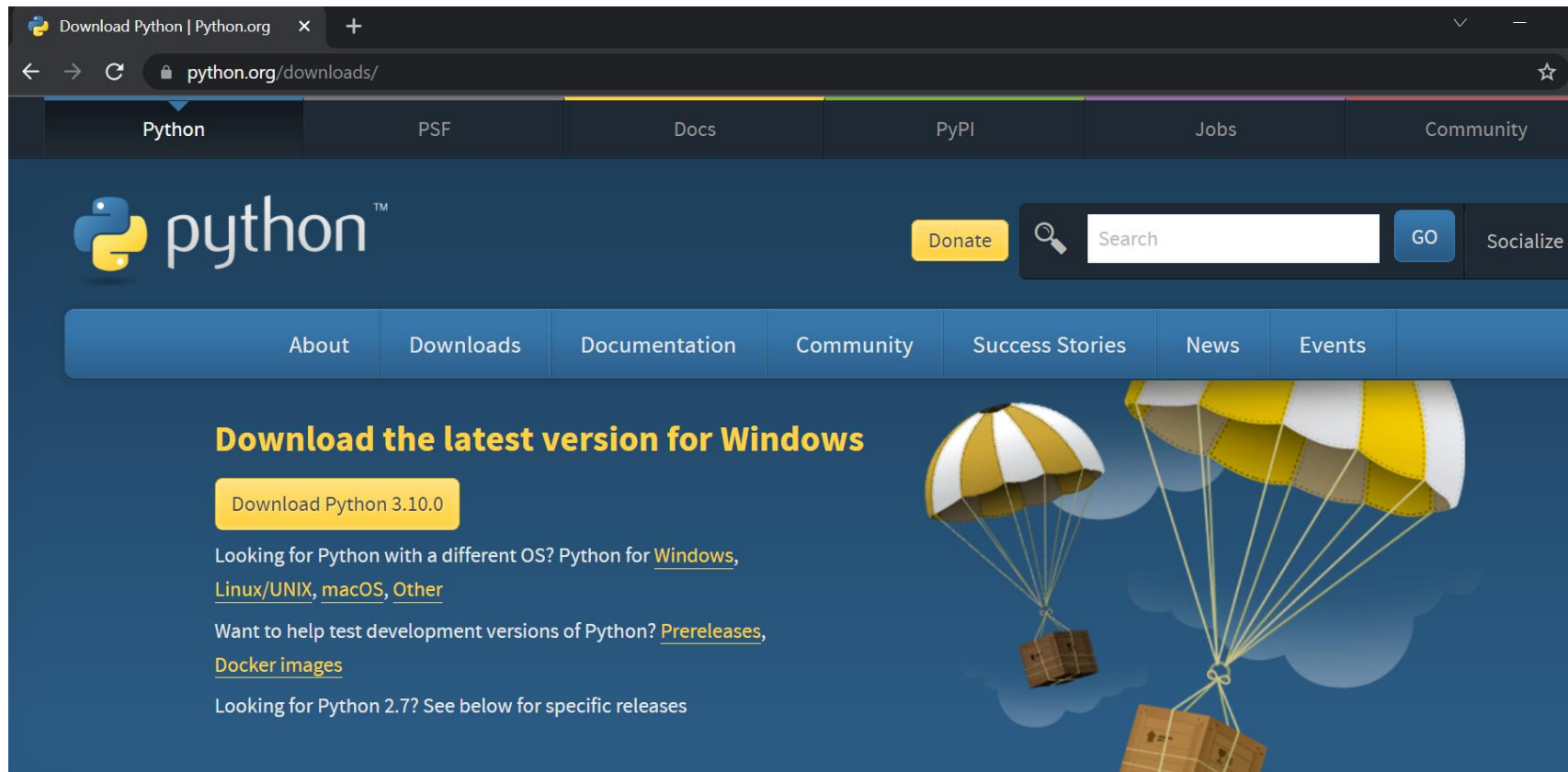




Installation For Windows users

Go to <https://www.python.org/downloads/>

(or google "python download") to reach this page:





Installation For Windows users

Download the installation file using this button: (version may vary, that's OK)

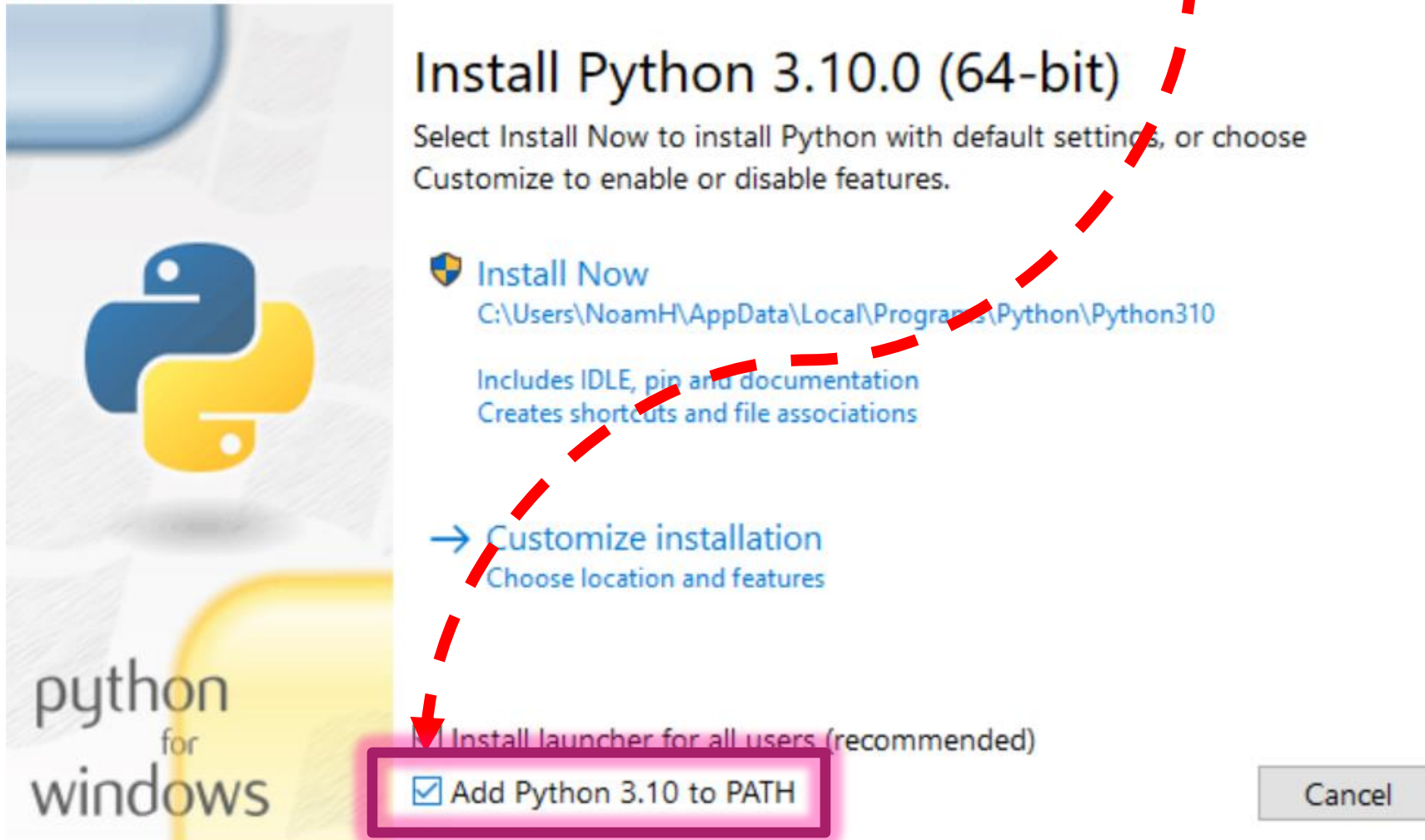
A screenshot of the Python.org website. The browser address bar shows 'python.org/downloads/'. The website has a dark blue header with navigation links: Python, PSF, Docs, PyPI, Jobs, and Community. Below the header is a blue banner with the Python logo, a 'Donate' button, a search bar, and a 'Socialize' link. A secondary navigation bar contains links: About, Downloads, Documentation, Community, Success Stories, News, and Events. A red dashed arrow points from the 'Downloads' link to a yellow button labeled 'Download Python 3.10.0'. This button is highlighted with a pink rectangular box. Below the button, there is text: 'Looking for Python with a different OS? Python for [Windows](#), [Linux/UNIX](#), [macOS](#), [Other](#)'. Further down, it says 'Want to help test development versions of Python? [Prereleases](#), [Docker images](#)'. At the bottom, it says 'Looking for Python 2.7? See below for specific releases'. The background of the page features a blue sky with clouds and two parachutes carrying boxes.



Installation For Windows users

Run the downloaded installation file, be sure to check this checkbox:

Python 3.10.0 (64-bit) Setup





Installation For Windows users

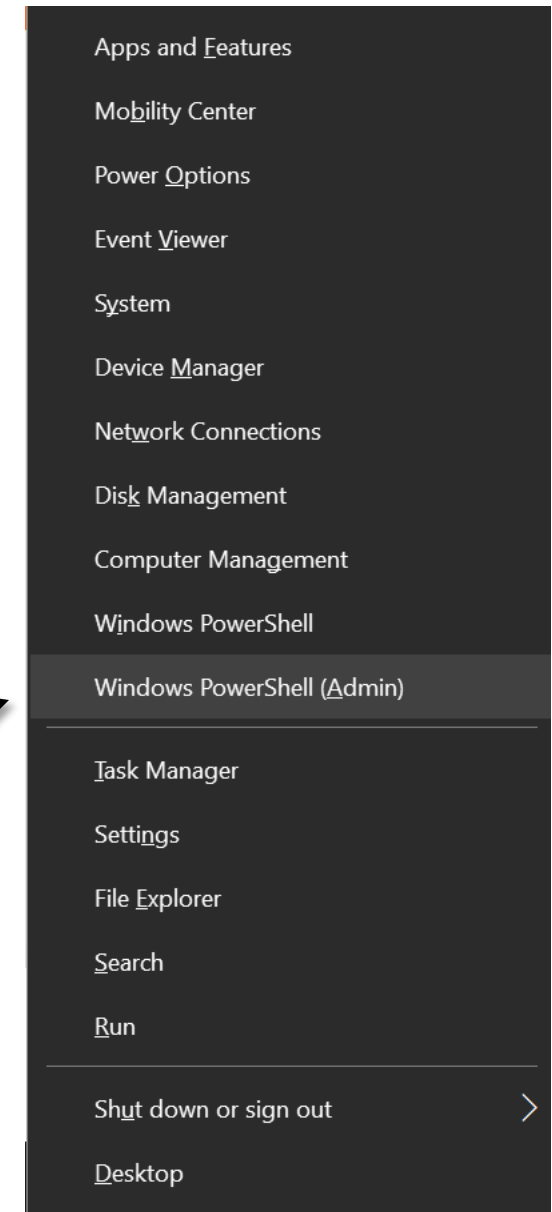
Now we need to install Window's subsystem for Linux.

Press the Windows key on your keyboard + the X key



Select "Windows PowerShell (Admin)".

This will open "PowerShell" in a new blue window.





Installation For Windows users

Copy this line into PowerShell:

```
pip3 install pandas dash lxml html5lib
```

```
C:\WINDOWS\system32> pip3 install pandas dash lxml html5lib
```

Then press enter. This will install needed python packages.



Installation For Windows users

In PowerShell, type:

```
wsl --install
```

```
PS C:\WINDOWS\system32> wsl --install
```

Then press enter and install WSL (Windows Subsystem for Linux).

After installation is completed, you will need to restart your computer.

Come back to the next slide after you finished restarting, I am waiting!



Installation For Windows users

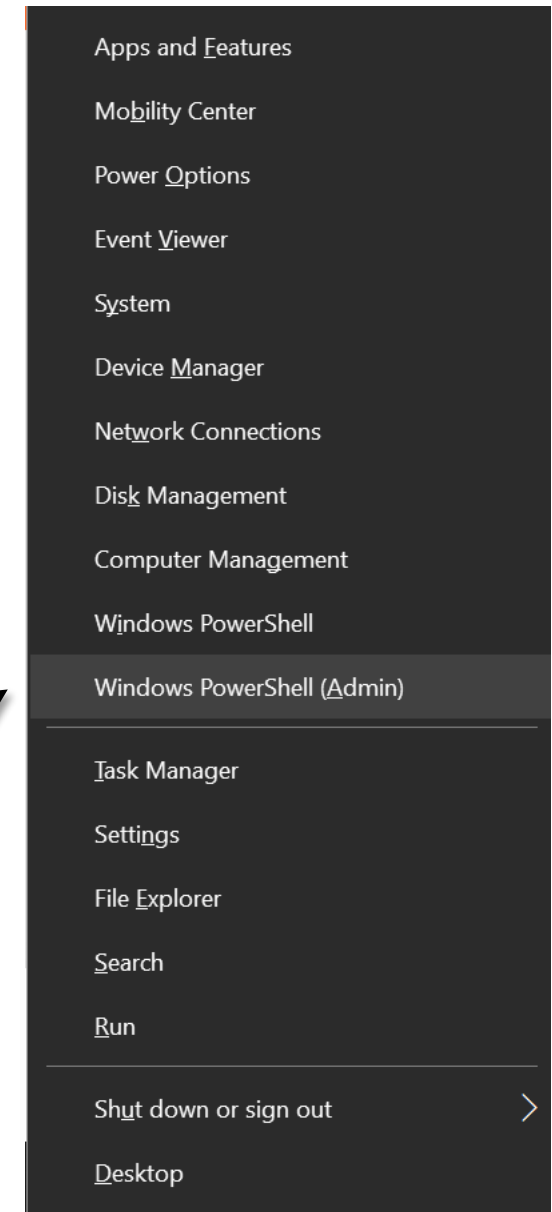
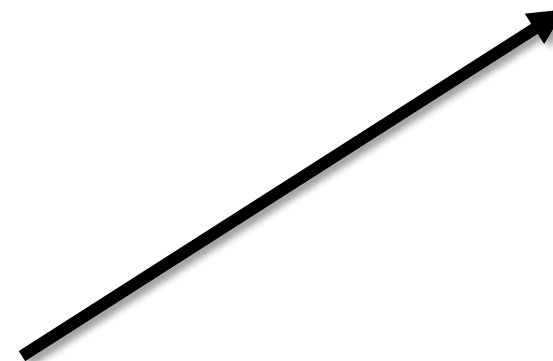
Now we need to install Linux and samtools.

So again:

Press the Windows key on your keyboard + the X key.



Select "Windows PowerShell (Admin)".





Installation For Windows users

In PowerShell type:

ubuntu

and press Enter. That will install ubuntu, a Linux environment. You'll be prompted to choose a username and a password – please remember them for the next step.

```
PS C:\WINDOWS\system32> ubuntu
```

Then type:

sudo apt-get update

and press Enter, you'll probably need to enter your password.

```
PS C:\WINDOWS\system32> sudo apt-get update
```



Installation For Windows users

Finally, type:

Sudo apt-get install samtools

and press Enter. This will install samtools, a software written for Linux that can analyze BAM files.

```
PS C:\WINDOWS\system32> sudo apt-get install samtools
```

That's it for installations! We are ready to start dealing with the good stuff.



Preparing a batch for analysis

Gather all *.bam* and *.bai* files of your input samples into a single folder.

All samples must have been sequenced by the same procedures (same sequencing machine, enrichment kit etc...) for reliable results.



Preparing a batch for analysis

Copy the "BAMdelbee-batch_preprocessor.py" python script into that folder, then start the script. This will prepare the batch for analysis – it can take several hours.

When the process is done, a "rename me.zip" file will be generated.



Preparing a batch for analysis

Name the "rename me.zip" file as you wish (probably by the name of the batch) and put it in BAMdelbee's "assets" folder. You can then start "BAMdelbee-application.py" to analyze your batch.



Analysis

Run "BAMdelbee-application.py" to start the application.

Remember that "rename_me.zip" file that you renamed and then moved to the assets folder? it is now an available batch for analysis.

Choose a batch for analysis using this auto-complete drop-down menu:

Select batch:

☒ hg38 ☐ hg19



Analysis

Samples within the batch will appear. Select those you want to analyze.

The analysis will display homozygous deletions found that are found only in the selected samples, but not in the unselected ones. You can also analyze just one sample of course.

181119

- ☐ 16264
- ☒ 16406
- ☐ 16382
- ☐ 16381
- ☐ 16437
- ☐ 16472
- ☐ 16151
- ☐ 16377

Analyze



Analysis

After pressing the "Analyze" button and waiting for loading to finish, you'll get a table with homozygous deletions, such as this:

	Coordinates	~Size (bp)	Genes
<input type="radio"/>	8:12393300-12393500	200	DEFB109A, FAM66A
<input type="radio"/>	11:117986450-117986950	500	IL10RA
<input type="radio"/>	11:117988300-117988700	400	IL10RA
<input type="radio"/>	11:117989250-117989700	450	IL10RA
<input type="radio"/>	14:106359900-106360000	100	IGH, IGHV3-33
<input type="radio"/>	17:39045100-39045200	100	LRR37A11P
<input type="radio"/>	19:54242800-54242950	150	



Analysis

Clicking a deletion will provide you with additional information and links about it.

450

	Coordinates	~Size (bp)	Genes
<input type="radio"/>	8:12393300-12393500	200	DEFB109A, FAM66A
<input checked="" type="radio"/>	11:117986450-117986950	500	IL10RA
<input type="radio"/>	11:117988300-117988700	400	IL10RA
<input type="radio"/>	11:117989250-117989700	450	IL10RA
<input type="radio"/>	14:106359900-106360000	100	IGH, IGHV3-33
<input type="radio"/>	17:39045100-39045200	100	LRR37A11P
<input type="radio"/>	19:54242800-54242950	150	

[IGV UCSC gnomAD](#)

interleukin 10 receptor subunit alpha

The protein encoded by this gene is a receptor for interleukin 10. This protein is structurally related to interferon receptors. It has been shown to mediate the immunosuppressive signal of interleukin 10, and thus inhibits the synthesis of proinflammatory cytokines. This receptor is reported to promote survival of progenitor myeloid cells through the insulin receptor substrate-2/PI 3-kinase/AKT pathway. Activation of this receptor leads to tyrosine phosphorylation of JAK1 and TYK2 kinases. Two transcript variants, one protein-coding and the other not protein-coding, have been found for this gene. [provided by RefSeq, Jan 2009].



Analysis

To use the IGV link, you must have IGV installed:

<https://software.broadinstitute.org/software/igv/>

You should also have your BAM file loaded into IGV.



UCSC gnomAD

interleukin 10 receptor subunit alpha

The protein encoded by this gene is a receptor for interleukin 10. This protein is structurally related to interferon receptors. It has been shown to mediate the immunosuppressive signal of interleukin 10, and thus inhibits the synthesis of proinflammatory cytokines. This receptor is reported to promote survival of progenitor myeloid cells through the insulin receptor substrate-2/PI 3-kinase/AKT pathway. Activation of this receptor leads to tyrosine phosphorylation of JAK1 and TYK2 kinases. Two transcript variants, one protein-coding and the other not protein-coding, have been found for this gene. [provided by RefSeq, Jan 2009].



That's it! We can't wait to hear about your findings.

Contact information:

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