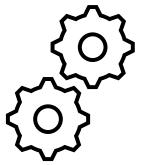
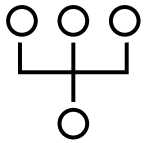
Instructions

Contents





1. Download and install Python (1/4)

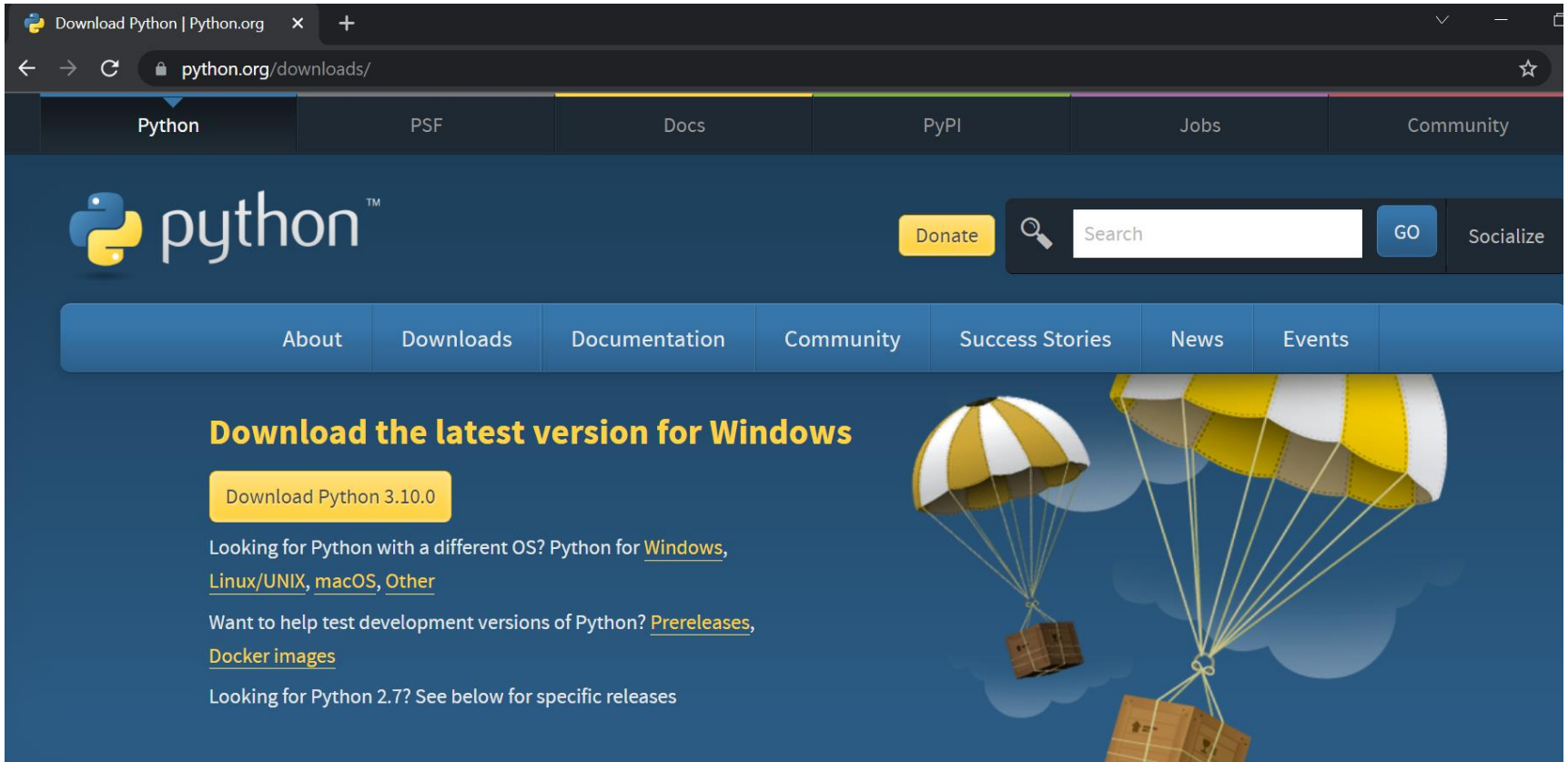
You need Python 3 and Pandas to run In-Silico Screener.

If you already have them installed, jump to page 10.

1. Download and install Python (2/4)

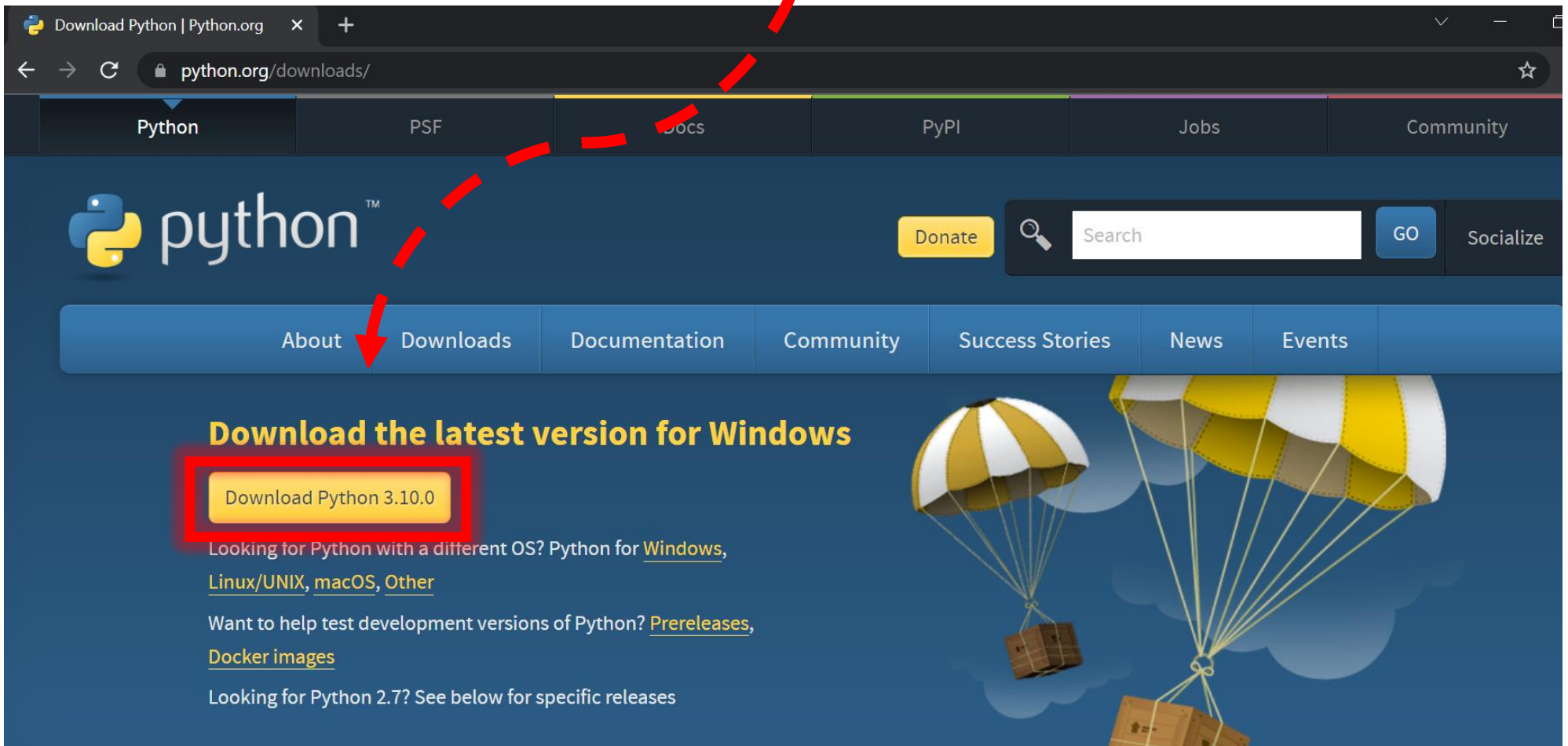
Go to **https://www.python.org/downloads/**

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



1. Download and install Python (3/4)

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



1. Download and install Python (4/4)

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

2. Installing Pandas – MacOS/Linux

Via terminal, install pandas using the command line:

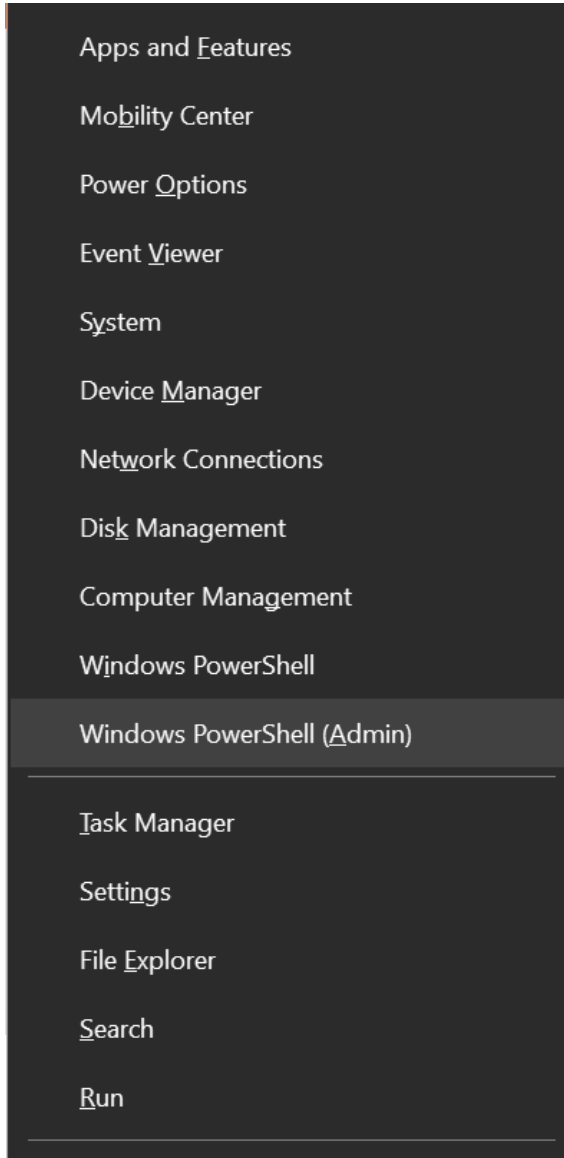
```
pip3 install pandas
```

2. Installing Pandas – Windows (1/2)

Press Windows key on your keyboard together with

the X key:

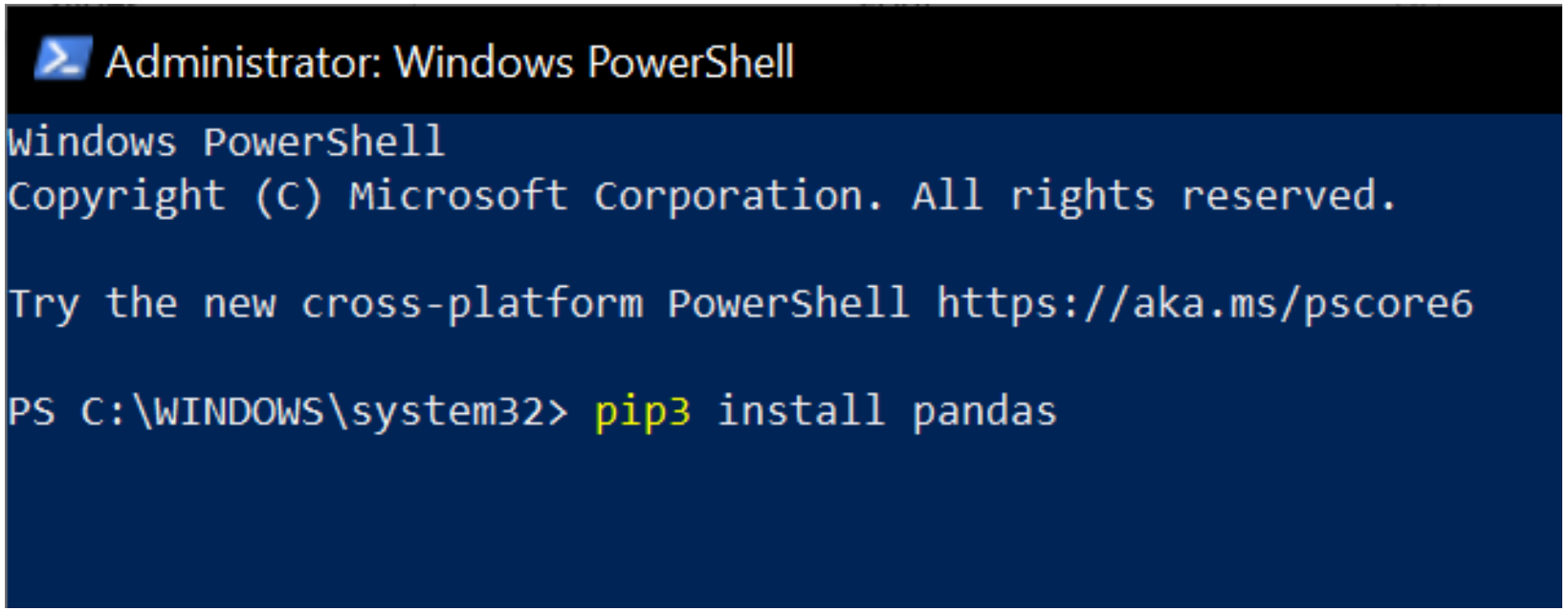
Select “Windows PowerShell (Admin)”.



This will open “PowerShell” in a new blue window.

2. Installing Pandas – Windows (2/2)

Type “pip3 install pandas” and press enter.



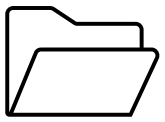
3. Download the python script using this button (on the website, not the one on this slide)

Download Python Script

4. Preprocessing (1/5)

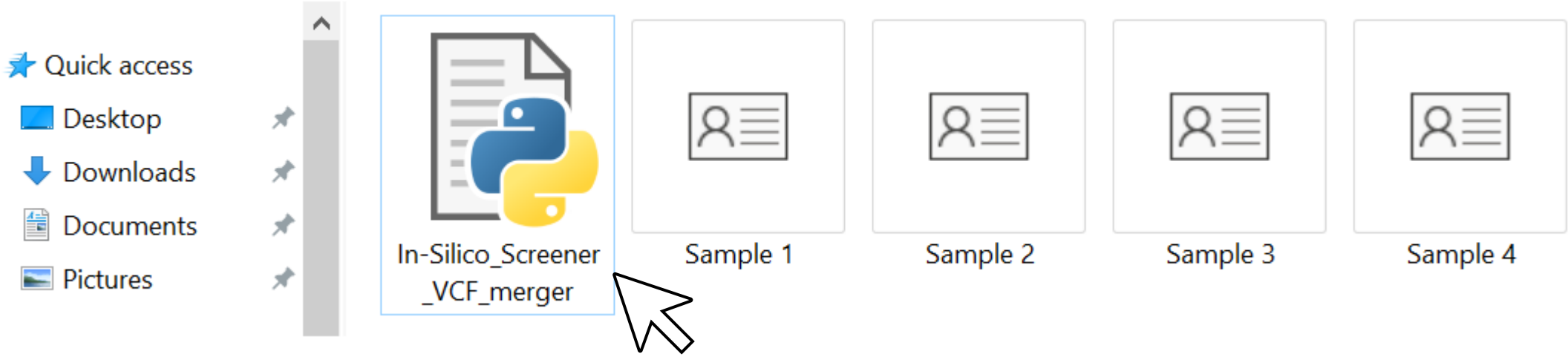
Place the python script from page 10 within a directory containing your whole-exome or whole-genome sequencing files in VCF formats (ending with either .vcf or .vcf.gz).

VCF files can be directly in the same folder, or within its sub-folders.



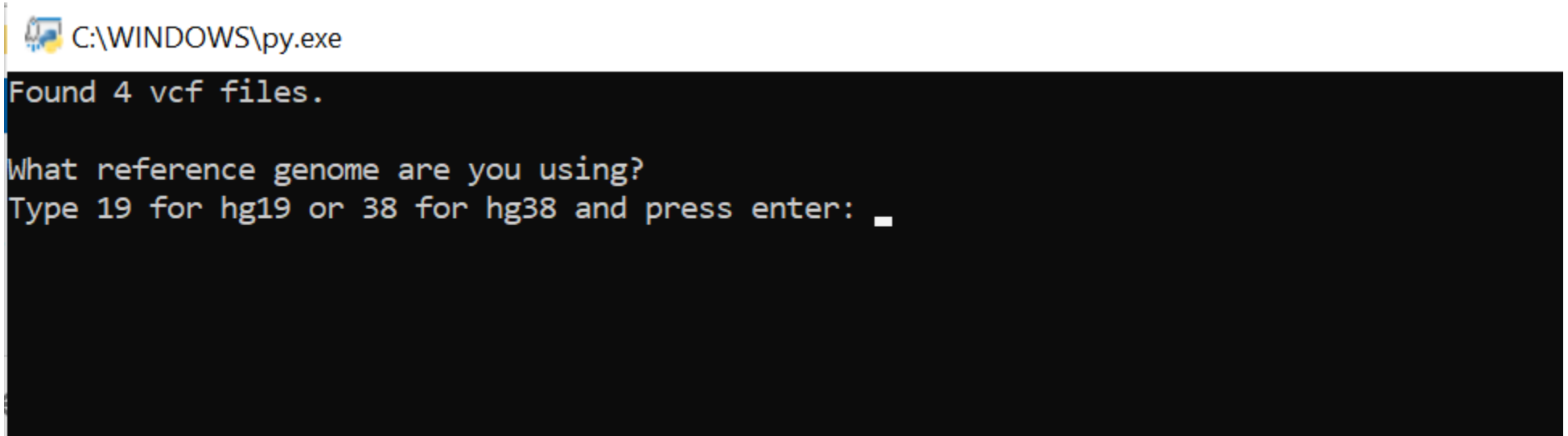
4. Preprocessing (2/5)

Activate the python script.



4. Preprocessing (3/5)

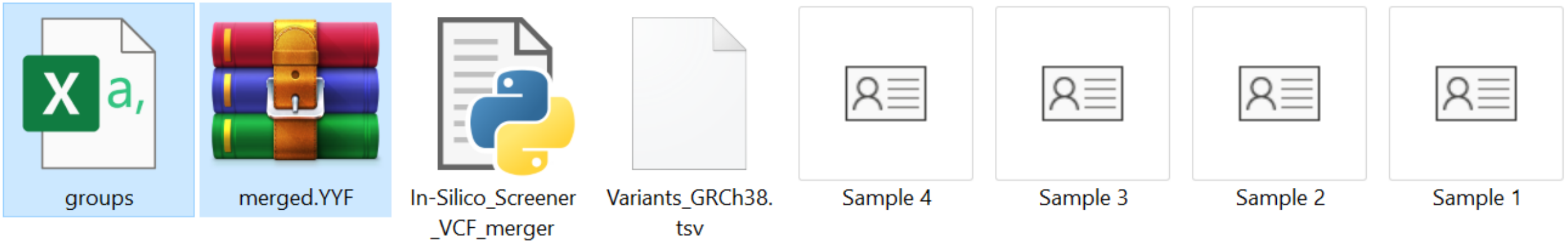
Follow the instructions, type “19” if you are using GRCh19/hg19 as reference genome, or “38” for GRCh38/hg38. Then press enter.



4. Preprocessing (4/5)

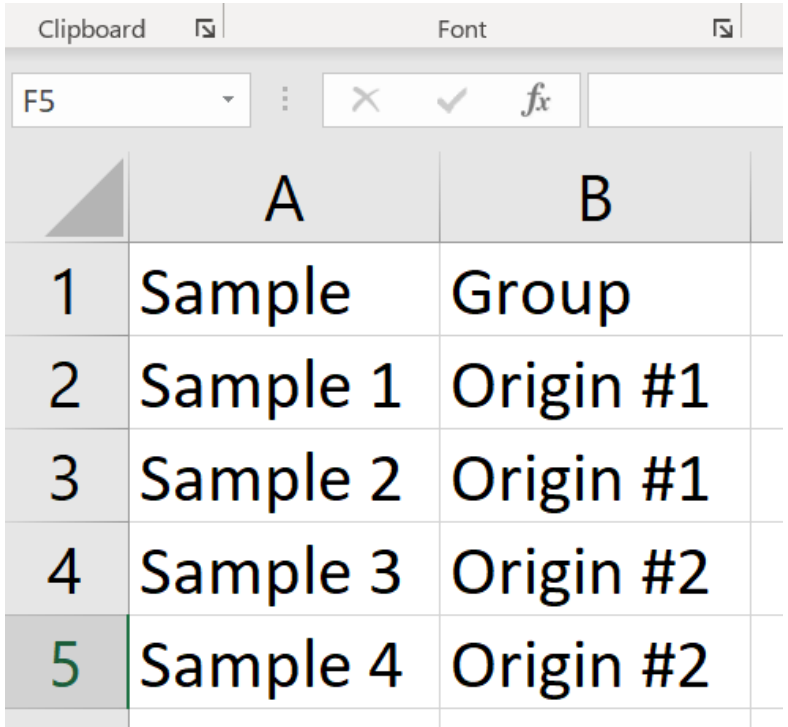
A variant database file will be downloaded. You can edit it and rerun to add additional variants.

Once the process is done, you will be provided with new files:



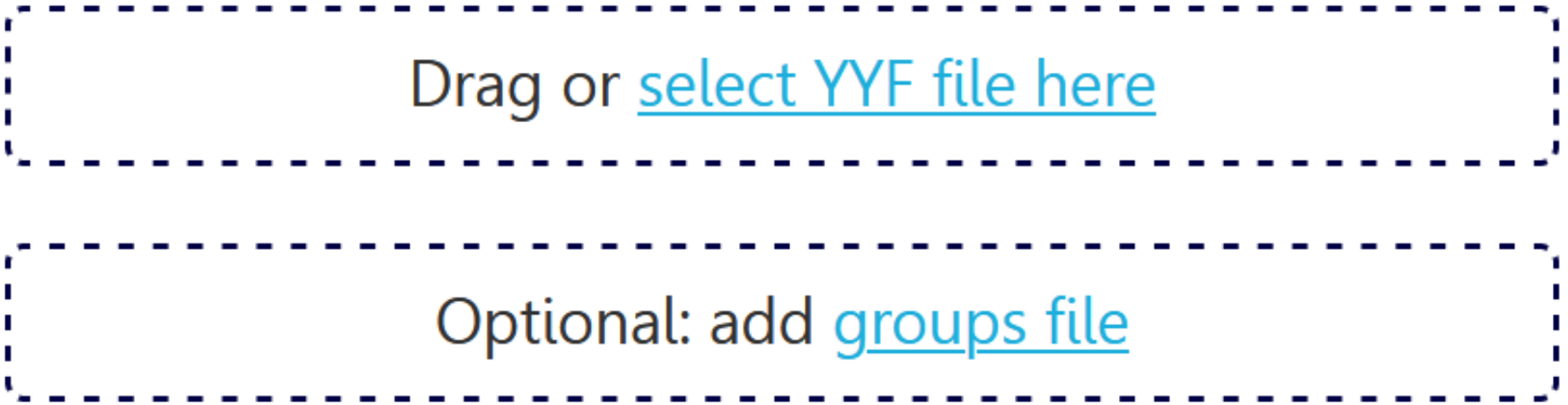
4. Preprocessing (5/5)

You can upload *merged.YYF* right away to begin an analysis, or tag samples by group name using the *groups* file.



5. Analysis (1/4)

Upload *merged.YYF* alone or with the groups file to the web-application:



Then press the Analyze button.

5. Analysis (2/4)

Pick a variant for further information

(heterozygotes, homozygotes and links)

OHAD BIRK'S LAB
THE MORRIS KAHN LABORATORY OF HUMAN GENETICS

In-Silico Screener

	Alleles	Phenotypes	Gene	Coordinates - hg38	Review Status
	filter data...				
○	1	Speech-language disorder 1	ZGRF1	4:112585555	no assertion criteria provided
○	1	Recombinase activating gene 2 deficiency;Primary immunodeficiency;Histiocy… medullary reticulosis	RAG2	11:36592849	criteria provided, single submitter
◉	1	not provided|Retinal dystrophy|Vitelliform macular dystrophy type 2	BEST1	11:61958159	criteria provided, multiple submitters, no conflicts

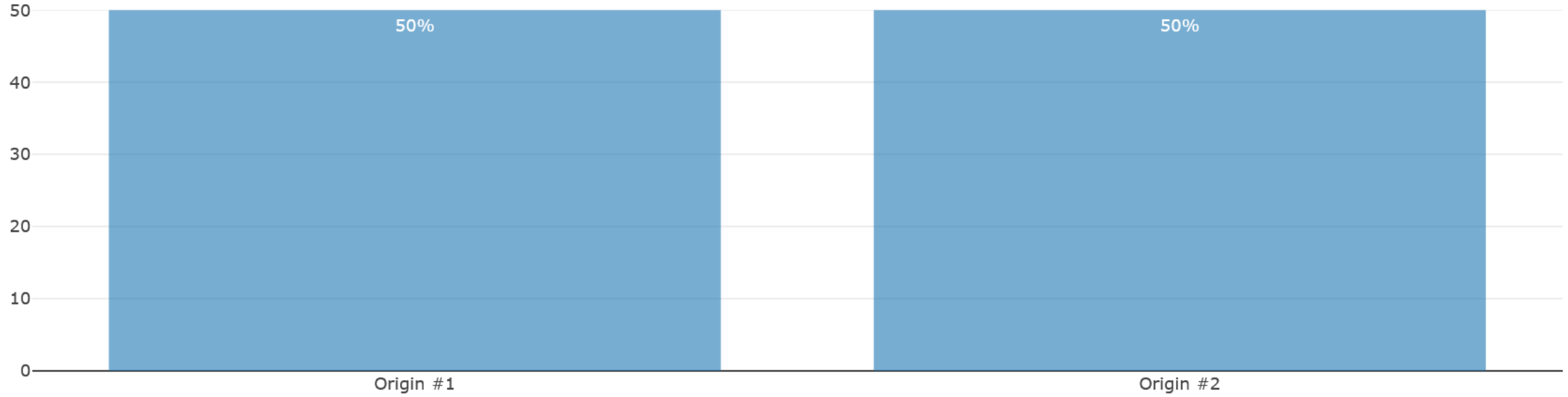
5. Analysis (3/4)

Be sure to review the variant's ClinVar, gnomAD and GeniePool page and frequency to further assess its pathogenicity.

ClinVar Page | gnomAD frequency | GeniePool

5. Analysis (4/4)

If you uploaded a *groups* file, you will be provided with additional information regarding variants' population distribution.



This is it.

Good luck!

In-Silico Screener