Tool Implementation Instruction

The ExonMutViz is implemented on Python3.x as well as it is made to run on Python 2.x. The dependent packages are: Matplotlib, Pillow (PIL.Image) and Numpy. (Note: This Tool will not run on Cygwin from Windows. If have already installed python 2.x OR python 3.x as AND configure numpy, matplotlib and Pillow dependencies either in linux or in Windows, you can run the *ExonMutViz* directly from command line)

The "configure.sh" file include complete implementation of the tool including. By this, user do not have to worry about calling the ExonMutViz manually. The scrip automatically takes care of:

- Configure Conda
- Configure Python3 with separate environment through the Conda
- Call the code automatically, generate, and store outputs in directory wherever user wants.

(See at the end detail of the three steps in Tutorial section).

Note: If the tutorial instruction fails somehow for the configuration of python, please do the manual configuration as explained in "Manual Configuration section" section in the end

Directory Structure

```
--- ExonMutViz
---- input
---- input_data.txt
---- reference_gene_models.txt
---- PapGene_SeqCovViz_Instructions.doc
---- output
----- (All sample-wise output results)
---- script
---- ExonMutViz.py (executable python script)
```

Specification of the Output

- The output of the tool generates individual PDF files; each represent a sample (The filename is corresponding to the Sample-ID).
- Each transcripts visualization is shown with Gene-ID and Transcript-ID.
- Each blocks of exons on a transcript are represented with rectangle shape with arrow at the end of the blocks. Size of the block is related to exon length. The arrow direction on

the exon block indicates orientation of transcript on DNA strand. The 5'->3' is in forward and 3'->5' in reverse direction and order of exon number should also be considered accordingly. Orientation of the transcripts can also be seen by looking at end of transcript with text either 5' or 3' on left and right position.

- Position of the reads are marked with fixed size pillar mark over the corresponding exons. The position of reads either at upper-side, if the read is from positive strand (+), or at lower-side, if the read is from negative strand (-).
- Color of the reads indicates above or below to a cutoff for the templates values of the reads. For 0> and <200 template value, the reads are shown with red color and for >=200 template value, the reads are shown with green color.
- Marker for overlapped reads are merged and plotted as single marker. i.e.,

PG-MAT-DNA-0004-01	CDKN2A_029_50CDKN2A	NM_000077.4	CCDS6510.1	chr9
- +	21974652 21974761	1776		
PG-MAT-DNA-0004-01	CDKN2A_001_21CDKN2A	NM_000077.4	CCDS6510.1	chr9
- +	21974743 21974859	25		

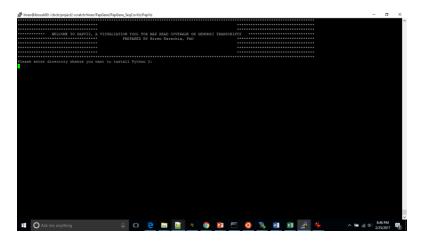
In above example of input file, the two reads whose mapping regions are partially overlapped. In such cases read markers are merged and considered as single mark. Thus reason of having less number of read marks coverage on a transcript as compared to the number of read entries in the input file is certainly because those overlapped reads are merged and plotted as single mark.

Delivering items

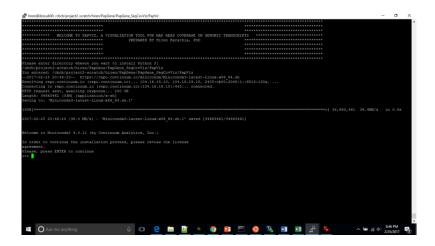
- 1) Code solution.
- 2) Instructions on how to execute your code solution (Microsoft Word).
- 3) The five output documents generated by your solution when applied to the supplied input file.

Tutorial of the configuration and run the tool

1. At running the "congfigure.sh" script on terminal by typing "bash configure.sh", you will see following screen. Here you will need to provide directory, where you uncompressed the ExonMutViz package i.e., type here "/home/John/ExonMutViz" if you uncompressed the ExonMutViz package inside the "/home/John" directory



2. After you entered your directory of ExonMutViz, first it will download Conda and will configure automatically. Next, it will configure python inside your directory. During this, it will ask few questions like accepting terms and conditions etc., you just need to press enter until you reach to a terminal where it will ask you the following:



It will ask you, as follow, for input directory where you wish to install miniconda3.

Miniconda3 will now be installed into this location:

<default directory>

- Press ENTER to confirm the location
- Press CTRL-C to abort the installation
- Or specify a different location below

Enter here whatever directory you entered in the first step ("/home/John/ExonMutViz") with adding "miniconda3" directory name i.e., enter here "/home/John/ExonMutViz /miniconda3"

At the end it will ask you followong

Do you wish the installer to prepend the Miniconda3 install location to PATH in your <home>/.bashrc? [yes|no]

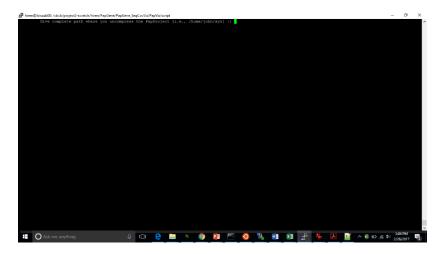
[no] >>>

Enter "no" here

Next it will ask you to install required software. Put there "yes"

>>> yes

At the end ExonMutViz Starts automatically. First, it will clear the screen and will ask you
to input directory where ever you store your ExonMutViz package. i.e., here you can
enter "/home/John/ExonMutViz"



Next it will ask you to insert directory where you want to store output i.e., you can enter "/home/John/ExonMutViz/output".

That's it, you will get your output stored in your selected directory Manual Configuration

If the above process fails, please follow manual configuration as follows. The reason of failing the above configuration may be if

- a. You are working with Ubuntu. In Ubuntu, I tested the configuration and came to know that there is permission issue that gives error in the automatic configuration.
- b. By mistake, you did not input the "miniconda3" directory correctly in step-2 above. If in the above step-2, you provide the "miniconda3" directory apart from where you uncompressed "ExonMutViz" folder, in next step it will not find that folder to configure dedicated Python folder, called "Py3" and at the end you will get error of configuration in mid and will not call the ExonMutViz program appropriately.
- c. Any other unknown reason!!!

But do not worry, just follow the steps, as given below and you will surely get the Python3 configuration and call the ExonMutViz correctly.

Steps of manual configuration and call the ExonMutViz

- Go to the folder where you uncompressed the ExonMutViz
 i.e., if you uncompressed the ExonMutViz inside "/home/John", go to ExonMutViz by
 typing
 - \$ cd /home/John/ExonMutViz
- 2. Type following command

\$ wget https://repo.continuum.io/miniconda/Miniconda3-latest-Linux-x86_64.sh

3. Next type

\$ bash Miniconda3-latest-Linux-x86 64.sh

You will follow self-explanatory instructions like

Please, press ENTER to continue

>>:

Just enter here, and keep entering until you don't reach to following

Do you approve the license terms? [yes|no]

>>>

Please answer 'yes' or 'no':

Type here "yes"

In following step you will be asked following

Miniconda3 will now be installed into this location: /home/hiren/miniconda3

- Press ENTER to confirm the location
- Press CTRL-C to abort the installation
- Or specify a different location below

[/home/<user>/miniconda3] >>>

If you press "ENTER", as it explained, by default the *miniconda3* will get configured in the /home/<user> directory, if not, you can input your desired directory where you want to configure the "*miniconda3*". In case of the second option, we recommend to input here your ExonMutViz directory i.e., /home/John/ExonMutViz/miniconda3

Next, you will be asked following:

Do you wish the installer to prepend the Miniconda3 install location to PATH in your /home/<user>/.bashrc ? [yes|no] [no] >>>

You can type either "yes" or "no", if you type yes, it will configure this python in your environment.

4. If you are inside "/home/John/ExonMutViz" and if you installed the miniconda3 from the previous step in the "/home/John/ExonMutViz" then type as follows instructions

If you are not using Ubuntu, you can do as follows

\$ /home/John/ ExonMutViz/miniconda3/bin/conda create --name Py3 python=3 numpy matplotlib Pillow

If you are using ubuntu then

\$ sudo /home/John/ExonMutViz/miniconda3/bin/conda create --name Py3 python=3 numpy matplotlib Pillow

\$ source activate Py3

If it doesn't work, then type

\$ source activate /home/John/ExonMutViz/miniconda3/envs/Py3

5. Next call the *ExonMutViz* as follows. Assume you are inside *ExonMutViz* directory i.e., "/home/John/ExonMutViz"

\$ python script/ExonMutViz.py

In the terminal, you will be asked to input the directory where you uncompressed the *ExonMutViz*. So, give complete directory path i.e., in our case "/home/John/ ExonMutViz"

Next, it will ask you to input desired output directory, you can put any directory you wish. In this case, you can put "/home/John/ExonMutViz /output"

AND

That's it, you will receive a message saying you can see your output inside the directory you mentioned. So easy!!! Isn't it??

Enjoy....

Wait, unfortunately 🕾

If through any unknown factor, still you get trouble in running the *ExonMutViz* or you find any bug in output of it, please write me on hirkarathia@gmail.com, I will be very much happy to help you.

THANK YOU