## **Assignment-1**

1. In your DSG docker folder, make a soft link for fastq file of RNA-seq profile

Using commans In -s /storage/vibhor/RNA-seq/SRR16292072.fastq.gz

a. Align it to hg19 genome using Tophat and STAR aligner tools

The indexes for Tophat are in /storage/vibhor/genomes/hg19/hg19/

The indexes for STAR aligner are at /storage/vibhor/genomes/hg19/starIndex

the gtf file for genes is here /storage/vibhor/genomes/hg19/hg19-Gencode.gtf

- b. After making bam files with aligner tools run cufflink to get the FPKM based expression of genes ( see http://cole-trapnell-lab.github.io/cufflinks/)
- c. Run htseq-count to get the read count.
- d. Plot the distribution of FPKM of genes in one sample. Tell what kind of distribution it fits best. Give argument/justification for your answer e. run cufflink to discover novel transcripts and promoters.

Write all the commands in the answer sheet and write a report on the stats like Total number of reads, number of reads with alignment.

Compare STAR aligner and Tophat in terms of speed and alignment percentage.

Note: first use command on the docker for the course, first type \$bash then type \$ source /storage/vibhor/addpath.sh to get access to different pre-installed tools, use tophat2

- 2. a)Make derivations and show relationship between binomial distribution and Poisson distribution
- b) Make derivation and show relationship between negative binomial distribution and Poisson distribution
- c) Make derivation for variance of poisson distribution
- d) Make derivation for variance of negative binomial distribution
- 3. You are given a set of numbers, assumed to be from a population of values following Poisson distribution

Now find the likelihood and p-value for occurrence of a number 6, given Possion distribution corresponding the above numbers.

Similarly do it for numbers, 7 and 8.

- 4. What is central limit theorem, give 2 examples where it helps in analysis in genomic data-science.
- 5. What is the 3-prime bias in RNA-seq data? How it could be created?
- 6. What could be the limitations of bridge PCR methods that are not present in emulsion PCR?
- 7. What is the difference between MAPQ and CIGAR fields in sam/bam file?