PBJ-1603220001050100 Seat No. _____

B. Sc. Bioinformatics (Sem. V) (CBCS) Examination

November / December - 2018

BI - 501 : Genomics

(New Course)

Time : $2\frac{1}{2}$ Hours] [Total Marks : **70**

Instructions :

(1) All questions are compulsory.

(2) The right side figures indicate total marks of the question.

1 Attempt the following : **14**

(a) Answer the following short questions : (all compulsory) **4**

(1) Why need to classify individual genes into families ?

(2) What are the limitations of classifications of individual genes into families ?

(3) Do alternative gene transcripts or splice variants have approved symbols ?

(4) Why can't punctuation be used in a gene symbol ?

(b) Answer any **one** of the following short questions : **2**

(1) What is GO and GOC ?

(2) What are the guidelines for naming in HGNC ?

(c) Answer any **one** of the following questions. **3**

(1) What are the three domains covered in gene ontology ?

(2) Principle of genome sequencing and assembly.

(d) Explain any **one** of the following questions in detail : **5**

(1) Explain in detail the three domains of GO Annotation.

(2) What are the processes involved in formation of Gene Family ? Explain each process in detail.

2 Attempt the following : **14**

(a) Answer the following short questions : **4**
(all compulsory)

(1) What is a Contig ?

(2) Which sequencing technique has the least error rate ?

(3) Which two key features on which SMRT is built ?

(4) Which are the sequencing methods based on Synthesis ?

(b) Attempt any **one** of the following short questions : **2**

(1) Mention the advantages and disadvantages of Biological Nanopore sequencing.

(2) What is Optical Mapping ? Explain in brief.

(c) Answer any **one** of the following short questions : **3**

(1) Explain the sequencing method which is based on light detection chain reaction.

(2) What are the advantages of SMRT ?

(d) Explain any **one** of the following question in detail : **5**

(1) Enlist various sequencing techniques and explain any one in detail.

(2) Provide the sequencing chemistry of DNA sequencing technique based on reversible termination.

3 Attempt the following : **14**

(a) Answer the following short questions : (all compulsory) **4**

(1) _____ format was developed to incorporate the phred-scaled base quality scores to facilitate the assessment of sequence quality.

(2) The complexity of sequence assembly is driven by _____.

(3) Define Exome.

(4) Name Categories of Exome Capture Technology.

(b) Answer any **one** of the following short questions : **2**

(1) What is the purpose of whole exome sequencing ?

(2) List out the advantages of exome sequencing.

(c) Answer any **one** of the following short questions : **3**

(1) Explain Identification of New Genetic Markers in Exome Sequencing.

(2) Explain SNP detection for DNA assembly.

(d) Explain any **one** of the following questions in detail : **5**

(1) Discuss cloud base solution for exome sequencing.

(2) Explain gene prioritization for interpretation of exome data.

4 Attempt following : **14**

(a) Answer the following short questions : **4**
(all compulsory)

(1) What is the function of Gene Prediction with Genetic Algorithm ?

(2) Give steps for genome annotation.

(3) Name the methods for measuring the abundance of transcripts.

(4) The Ensemble gene annotation process can be divided into which four main phases ?

(b) Answer any **one** of the following short questions : **2**

(1) What is NCBI Prokaryotic Genome Annotation Pipeline ?

(2) Briefly write about the methods implemented in KAAS.

(c) Answer any **one** of the following short questions : **3**

(1) What is an optimized approach for annotation of large eukaryotic genomic sequences using genetic algorithm ?

(2) How will you select the target and reference sequences ?

(d) Explain any **one** of the following questions in detail : **5**

(1) The Ensemble gene annotation system.

(2) Protein-coding model building.

5 Attempt the following : **14**

(a) Answer the following short questions : **4**
(all compulsory)

(1) What are the two main types of Pseudogenes ?

(2) SNP density can be predicted by the presence of _____.

(3) What is the principle of CPIC ?

(4) What are the two most characterized epigenetic modifications ?

(b) Answer any **one** of the following short questions : **2**

(1) Explain processed pseudogene.

(2) What are the duplicated pseudogenes ?

(c) Answer any **one** of the following questions. **3**

(1) How are the pseudogenes predicted ?

(2) In all types of SNPs either it would have observable phenotype or it results into disease. Explain the sentence.

(d) Explain any **one** of the following questions in detail. **5**

(1) Which methods were used earlier based on principle of personalized medicine ? Explain.

(2) Explain PharmGKB.
