

## **Guided Self Learning (GSL) 2 – GENETICS**

1. Define medical genetics.
2. What is Central Dogma of molecular biology?
3. Describe the difference between DNA and RNA
4. Define mutation. Explain the following terminologies:
  - i. missense mutation
  - ii. nonsense mutation
  - iii. synonymous mutation
  - iv. inframe mutation
  - v. frameshift mutation
5. List down 5 examples of single gene disorders.
6. What is cytogenetics?
7. Explain the symbols used in describing a karyotype.
  - i. p
  - ii. q
  - iii. cen
  - iv. del
  - v. dup
  - vi. inv
  - vii. t
8. Give the karyotype patterns of the following:
  - i. Edwards syndrome
  - ii. Patau syndrome
  - iii. Klinefelter syndrome
  - iv. Turner syndrome
  - v. Down syndrome
9. Define pedigree. What are the general symbols used for pedigree drawing? Prepare your own family pedigree for at least 3 generations.
10. Describe the Mendelian Laws of Inheritance.

*Please answer all the questions. If you have any enquiry, please contact lecturers from Human Genome Centre who are involved in GMT102. Thank you.*