

GSL 2 – GENETICS (1 DECEMBER 2020)

1. Define medical genetics.
2. Define three (3) principles of Mendelian Inheritance.
3. Define genetic code. Identify the start and stop codon.
4. Define mutation. Explain the following terminologies:
 - i. missense mutation
 - ii. nonsense mutation
 - iii. synonymous mutation
 - iv. inframe mutation
 - v. frameshift mutation
5. Describe the differences between gene polymorphism and gene mutation
6. List down 5 examples of single gene disorders.
7. What is cytogenetics?
8. Explain the symbols used in describing a karyotype.
 - i. p
 - ii. q
 - iii. cen
 - iv. del
 - v. dup
 - vi. inv
 - vii. t
9. Define syndrome. Give the karyotype patterns and clinical features of the three (3) common autosomal related syndromes and two (2) sex chromosomes related syndrome
10. Define pedigree. What are the general symbols used for pedigree drawing? Prepare your own family pedigree for at least 3 generations.

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