University of the Philippines Manila
COLLEGE OF MEDICINE

Master of Science in Genetic Counseling Program

GC 201- Principles of Genetic Counseling
1ST LONG EXAMINATION

Name:_____Student No. _____

Direction: Answer all the questions <u>briefly but comprehensively</u>. Please submit through the Virtual Learning Environment in PDF format before November 10, 11:59pm.

1) Discuss how the focus of genetic counseling shifted from its inception in mid-1900s to the present. (10 points)

2) You are providing genetic counseling to a patient diagnosed with Maple Syrup Urine Disease (MSUD). MSUD has an autosomal recessive inheritance pattern. Please explain –

   a) What is autosomal recessive inheritance? (5 pts)

   b) What is the recurrence risk for your patient's parents to have another child diagnosed with MSUD? (5 pts)

   c) Using the Reciprocal Engagement Model of Genetic Counseling, how would you approach the family in providing genetic counseling? (15points).

3.) You are providing genetic counseling to a family with a child diagnosed with G6PD deficiency, an x-linked recessive disorder. In counseling the patient's family, you are guided by the genetic counseling tenet that genetic information is key and important to share. What specific genetic information would you share to this patient's family? (10 points).

4) A 25 year-old married woman (Mrs. Lloyd) came to your clinic to discuss her risk given her family history of X-linked spinal and bulbar muscular dystrophy (SBMA or Kennedy's disease). You took the family history and you found out that she has one affected brother with SBMA, age 18 years old.

   Mrs. Lloyd has two unaffected sons, ages 5 and 8 years of age. Her parents are healthy and living. The family is of Filipino ancestry and there is no reported possibility of consanguinity.

   a. Draw the complete family pedigree and appropriately indicate the individuals who are affected or unaffected, as well as use an arrow to indicate the consultand. (5 points)

   b. What is the PRIOR risk that Mrs. Lloyd is a carrier for SBMA? (5 points)

c. Using Bayesian analysis, what is the probability that Mrs. Lloyd (your patient) is a carrier for SBMA given that she already has 2 unaffected sons? (5 points)

d. Using the carrier risk estimate following the Bayesian analysis, what is the Mrs. Lloyd's risk to have a son in her next pregnancy affected with SBMA? (5 points)

5) You are seeing a patient newly diagnosed with congenital adrenal hyperplasia, an autosomal recessive disorder screened through the newborn screening. The family was shocked upon learning of the patient's condition and started to ask questions. Using the meaning-making model in genetic counseling

a. How would you provide support to this patient's family? Give concrete examples of interventions (5 points)

b. How can you facilitate empowerment of the patient's family? Give concrete examples of interventions (5 points)

6) A family is affected with Huntington disease, an adult-onset neurologic disorder. Fifty percent (60%) of people who inherit the HD gene show some symptoms of HD by the age of 65 years. John is 65 years old and does not show any HD symptoms, but his father died of HD. What is the probability that John's son, Bob, inherited the HD gene?

a. Draw the pedigree (5 pts)

b. Using Bayesian analysis, what is John's risk to have the HD gene (5 pts)?

c. What is John's son's risk to have the HD gene (5 pts)?