EXAMPLE QUESTIONS FROM THE ABGC CERTIFICATION EXAMINATION

1. Which of the following features identified by fetal ultrasound is **MORE** commonly associated with trisomy 18 than with trisomy 21?

A. nuchal thickening
B. duodenal atresia
C. echogenic bowel
D. choroid plexus cyst

2. A 20-year-old man and his family are seen for genetic counseling to discuss his recent diagnosis of Charcot-Marie-Tooth (CMT) disease. After introductions, which of the following would be the **BEST** next step for the genetic counselor to take?

A. explain the recurrence risks of CMT
B. ask them what they have been told about CMT
C. collect a family and medical history
D. review the results of his genetic testing

3. A 30-year-old woman who had a previous stillbirth comes for prenatal genetic counseling because her serum screening results show a very low estriol with relatively normal levels of AFP, hCG, and inhibin. To help determine the **MOST** likely diagnosis for this pregnancy, the genetic counselor should inquire about which of the following abnormalities in the stillborn child?

A. hypospadias and congenital heart defect
B. hydrocephalus and adducted thumbs
C. absent radii and cystic kidneys
D. short limbs and polydactyly

4. A 35-year-old woman comes for genetic counseling because of a family history of polycystic kidney disease (PKD) consistent with autosomal dominant inheritance and confirmed by review of medical records. Genetic testing has not been performed. The woman's renal ultrasound showed a single unilateral kidney cyst. The patient states, "Now that I have PKD, my children are at 50% risk to have it, too. They need to have DNA testing." Which of the following is the **BEST** response to the patient's statement?

A. agree that she meets diagnostic criteria and recommend her children have DNA testing
B. agree that she meets diagnostic criteria and recommend her children have renal ultrasounds
C. explain that she does not meet diagnostic criteria and recommend an affected relative have DNA testing
D. explain that she does not meet diagnostic criteria and recommend she have DNA testing

5. A 12-year-old girl is referred to the genetics clinic because of a childhood history of bilateral retinoblastoma. Due to her treatment in infancy with bilateral external beam radiotherapy, which of the following additional cancers is she **MOST** likely to develop?

A. breast cancer
B. leukemia
C. osteosarcoma
D. pineal blastoma

6. Which of the following techniques would be used to detect a previously identified germline *APC* mutation in an unaffected first-degree relative?

A. site-specific sequencing
B. full gene sequencing
C. heteroduplex analysis
D. protein truncation assay

7. A 27-year-old woman who is 9 weeks pregnant has a nephew with Down syndrome. The woman is concerned about the risk to her current fetus. Which of the following is the **BEST** first step in counseling this woman about her risk?

A. karyotype her to determine whether she has a translocation
B. offer her prenatal diagnosis
C. recommend she have serum screening and fetal ultrasound
D. request a copy of the nephew's karyotype

8. A 35-year-old woman, recently diagnosed with an invasive ductal carcinoma of the breast, comes for genetic counseling with her 30-year-old sister. Their mother died of breast cancer at age 52 and was the only other affected relative. To determine the risk to her daughter, the woman has comprehensive *BRCA1* and *BRCA2* genetic testing with negative results. Which of the following statements by this patient should make the counselor **MOST** concerned that she does not fully understand these results?

A. "I am not at significantly increased risk for ovarian cancer."
B. "My daughter will not need to have *BRCA* testing."
C. "My sister is not at increased risk for breast cancer."
D. "I am considering bilateral mastectomy."

9. A genetic counselor meets with the parents of a 15-year-old girl who was recently diagnosed with Turner syndrome. The parents do not want their daughter to be informed about her diagnosis because they feel that it will upset her and they prefer to wait and tell her themselves when they think that she is ready. Which of the following is the **BEST** approach by the counselor?

A. discuss strategies for talking about this diagnosis with girls her age
B. encourage the parents to join the local Turner syndrome support group
C. honor the parents' request because the patient is a minor
D. insist they share the diagnosis with their daughter as soon as possible

10. A genetic counseling student has just seen a couple whose first-trimester screening result showed a 1 in 5 chance for the fetus to have trisomy 18. During the session, the husband made several disparaging comments about his wife and stated, "Of course she wants a CVS, she always worries about everything!" By the end of the session, the student was very angry with the husband. As she discusses the case with her supervisor, she realizes the husband reminded her of her own father. Which is the **BEST** approach for the supervisor to take with this student?

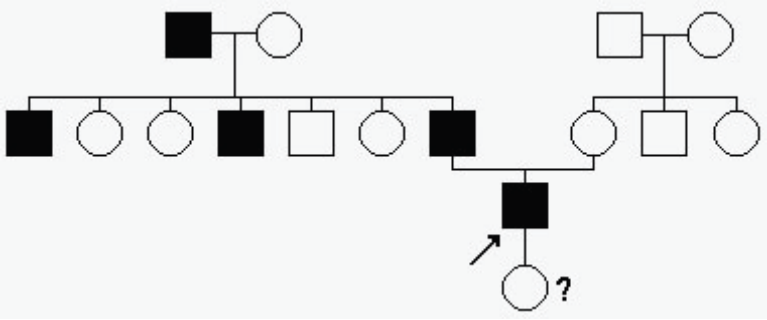
1. encourage her to engage in self-reflection.
2. help her identify the source of her feelings.
3. discuss with her ways to minimize transference.
4. share with her that this reaction is not unusual.

A. 1, 2, and 3 only
B. 1, 2, and 4 only
C. 1, 3, and 4 only
D. 2, 3, and 4 only

11. A 22 year old woman who is 20 weeks pregnant had a fetal ultrasound which showed spina bifida and slightly enlarged cerebral ventricles. After the genetic counselor discusses the possible diagnosis and the unpredictability of the outcome, the woman expresses her reluctance to terminate the pregnancy. Her husband remains very quiet and says that he will go along with his wife's decision. Which is the **BEST** approach for the genetic counselor to take?

A. support the woman's decision
B. engage the husband in the decision-making process
C. refer the couple for family therapy
D. suggest that the couple continue this discussion at home

12.



A trait has a population frequency of 1 in 40,000. Assuming the most likely mode of inheritance as shown in the pedigree, what is the probability that the proband's daughter has inherited the familial mutation?

A. 0
B. 1/100
C. 1/200
D. 1/2

KEY

1. D
2. B
3. A
4. C
5. C
6. A
7. D
8. C
9. A
10. B
11. B
12. D