

Practice problems Block IV

1. You are a pediatrician and notice that your patient, a 2-month old former 29 week premature infant has only had one newborn screening sample sent and it was marked as having been sent 2 days after a blood transfusion was given to the infant. You should consider which of the following?
 - A. Send a repeat sample immediately
 - B. Send a repeat sample 90 days after the last transfusion
 - C. Assume that the first screen was adequate
 - D. Just run plasma amino acids to rule out aminoacidopathy
 - E. Whole genome sequencing

Answer: B

2. Newborn screening is restricted to which **one** of the following?:
 - A. Disorders which will not be expressed until the age of 5 years old
 - B. Any disorder which will vary in ethnic or racial distribution
 - C. Any disorder that can be detected at birth
 - D. Disorders of chromosomal aneuploidy
 - E. Disorders which could be effectively treated

Answer: E

3. Which of the following is true of a highly sensitive screening test?
 - A. It will also be highly specific.
 - B. It has a higher risk of false positives.
 - C. It has a higher risk of false negatives.
 - D. It does not require a confirmatory test in cases of positive results.
 - E. More than one of the above is true.

Answer: B

4. Based on medical and family history, a physician suspects a patient has familial adenomatous polyposis. How can she confirm the diagnosis ?
 - A. Test for a mutation on the *APC* tumor suppressor gene on chromosome 5q
 - B. Test for a mutation on the *ABL/BCR* gene on chromosome 9
 - C. Test for a translocation between chromosome 9 and 22 [t(9;22)]
 - D. Test for a deletion of 13q14
 - E. Test for mutations in the HNPCC related genes

Answer is A

5. Mammalian DNA methyltransferase 1 (DNMT1)
- A. Is especially important for de novo methylation
 - B. Has the highest affinity for unmethylated CpG pairs
 - C. Is important for changing DNA methylation patterns
 - D. Is the main enzyme for maintaining DNA methylation patterns
 - E. Is not functional/active during early embryogenesis in mammals

Answer is D

6. Which of the following is not restricted while prescribing dietary therapy for a young infant with phenylketonuria (PKU)?
- A. Breast milk intake
 - B. Tyrosine intake
 - C. Phenylalanine intake
 - D. Protein intake

Answer is B