1. The three chromosomal abnormalities that cause Down syndrome are:
A. trisomy 21 ; translocation ; mosaicism
B. trisomy 23 ; translocation ; mosaicism
C. trisomy 21 ; translocation ; Fragile X
D. trisomy 23 ; translocation ; Fragile X
E. trisomy 23 ; mosaicism ; Fragile X
Ans: A

2. What is the most common cause of Down syndrome?
A. Paternal nondisjunction
B. Maternal translocations
C. Maternal nondisjunction
D. Paternal translocation
Ans: C

3. The enzyme that metabolizes 6-mercaptoputine is:
A. DPD.
B. TPMT.
C. CYP2D6.
D. NAT1.
Ans: B

4. A drug substrate for the metabolizing enzyme CYP2D6 is:
A. tamoxifen.
B. codeine.
C. diazepam.
D. irinotecan.
Ans: B
5. Why does a polymorphism in CYP2D6 decrease the efficacy of codeine?
A. Alters the number of AhR
B. Decreases uptake of codeine by the brain
C. Increases excretion of codeine
D. Increases Phase II metabolism
E. Slows formation of morphine
Ans: E

6. What would be the expected result when the area around an oncogene is hypomethylated?
A. Cell division increases; cancer risk increases.
B. Cell division increases; cancer risk decreases.
C. Cell division decreases; cancer risk decreases.
D. Cell division decreases; cancer risk increases.
Ans: A

7.Which two of the following metabolizing enzymes polymorphism is important
in proper warfarin dosing?
A. TPMT and CYP2D9
B. CYP2D9 and CYP2D19
C. VKORC1 and CYP2C9
D. NAT1 and VKORC1
Ans: C
8. The gene of the metabolizing enzyme that catalyzes the transfer of an acetyl
moiety from acetyl-CoA to hydralazine is:
A. CYP2C6.
B. CYP2D9.
C. NAT1.
D. UGT1A1.
Ans: C
9. Regarding Pharmacogenetics, a phase 1 enzyme metabolizes drugs by:
A. conjugating an acetyl group to a drug compound.
B. transferring a methyl group to a drug substrate.
C. conjugating an amine group to a drug substrate.
D. hydroxylating a drug substrate.
Ans: D
10. The HLA gene and its variants that have been associated most closely with drug
hypersensitivity and that can be assessed in a laboratory is HLA-:
A. A
B. B.
C. C.
D. D.
Ans: B

11. Which of the following represents a karyotype of a male with a balanced Roberstonian translocation of chromosome 13 and 14:
A. 46, XY, der (13;14) (p21:q13)
B. 46, XY, t (13;14) (q23:q22)
C. 45, XY, der (13;14) (p21:q13)
D. 45, XY, der (13;14) (q10:q10)
E. 46, XY, der (13;14) (q10:q10)
Ans: D

12. Which histone is replaced in nucleosomes found in the vicinity of centromeres?
A. histone H1
B. histones H2A
C. histone H2B
D. histone H3
E. histone H4
Ans: D

13. What is the relationship between the cause of Prader-Willi syndrome and the cause of Angelman syndrome?
A. Both involve chromosome 15; Prader-Willi involves a deletion of genetic material on this chromosome, while Angelman Syndrome involves extra material on this chromosome.
B. Both involve chromosome 15; Prader-Willi involves a deletion of genetic material from the father whereas Angelman involves a deletion of genetic material from the mother.
C. Both involve the deletion of genetic material; Prader-Willi from chromosome 15 and Angelman from chromosome 16.
D. Both involve nondisjunction; Prader-Willi on chromosome 15 and Angelman on chromosome 16
Ans: B

14. Hyperphagia is associated with which condition?
A. Down syndrome
B. Angelman syndrome
C. Williams syndrome
D. Prader-Willi syndrome
Ans: D

15. The field of Pharmacogenetics:
A. tailors drug treatments to individual genotypes.
B. tailors drug treatments to individual phenotypes.
C. prescribes generic treatments to individuals.
D. prescribes antibiotics to treat individual bacterial infections.
Ans: A

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