Exam Procedures:

STEP 1- NAME (*Print clearly*) _____ _____

(first) *(last)*

STEP 2 – Fill in your answer sheet, using a #2 scoring pencil, as follows:

- ☑ Your Student PID Number (excluding "A")
- ☑ Your last name and first name
- ☑ Course ID in "subject" …… **this is BMB 526 Exam #3**
- ☑ Date …… **12/12/13**
- ☑ Exam form in "period" …..**this is form A**
- ☑ By signing this coversheet for this exam, the student certifies that he/she has adhered to the policies of academic honesty in the performance of this exam.

_____

Signature

STEP 3 - Read these instructions:

- ☑ Make sure your exam has **60** questions.
- ☑ Read each question very carefully. Choose the single, best answer and mark this answer on your answer sheet. No points will be added for correct answers which appear on the exam page but not on the answer sheet.
- ☑ Cell phones must be off (not on vibrate) and stored with your bags/backpacks/other materials.
- ☑ The proctors have the authority/responsibility to assign any student a different seat at any time, without implication and without explanation, before or during the examination, as they deem necessary. Accomplish any relocation quietly and without discussion.
- ☑ We will not answer questions of clarification. However, if you think there is an error on your exam, summon an exam proctor.
- ☑ When you finish, place all exam materials (except the tear sheet) into the manila envelope. When you leave the exam room, please turn in your envelope to the proctors. Once you exit the auditorium, please leave the area. Hallway conversations disturb those still taking the exam.
- ☑ There will be answer keys to this exam posted on the course website by 5:00 p.m. the day of the exam. You may wish to copy your responses from your answer sheet onto the answer grid on the LAST page of this exam so that you can check your results. You can tear off the last page and take it with you.
- ☑ We will close the exam promptly at **11:00 a.m**. At the announcement of the examination end time, the examination and scantron and images (if provided as part of the examination) must immediately be placed into the manila envelope provided.

STEP 4 – Wait until instructed to proceed with the exam!

The information provided below may be useful in answering some questions.

The "Wobble" Rules of Codon-Anticodon Pairing	
5' Nucleotide of Anticodon	**3' Nucleotide of Codon**
C	G
A	U
U	A or G
G	C or U
I	U, C, or A
3' Nucleotide of Codon	**5' Nucleotide of Anticodon**
G	C, U
A	I, U
C	I, G
U	I, G, A

First position (5' end)	Second position U	C	A	G	Third position (3' end)
	Phe	Ser	Tyr	Cys	U
	Phe	Ser	Tyr	Cys	C
	Leu	Ser	Stop	Stop	A
U	Leu	Ser	Stop	Trp	G
	Leu	Pro	His	Arg	U
	Leu	Pro	His	Arg	C
	Leu	Pro	Gln	Arg	A
C	Leu	Pro	Gln	Arg	G
	Ile	Thr	Asn	Ser	U
	Ile	Thr	Asn	Ser	C
	Ile	Thr	Lys	Arg	A
A	Met	Thr	Lys	Arg	G
	Val	Ala	Asp	Gly	U
	Val	Ala	Asp	Gly	C
	Val	Ala	Glu	Gly	A
G	Val	Ala	Glu	Gly	G

INFORMATION ON COMPONENTS OF RIBOSOMES

I. Prokaryotes (e.g. *E. coli*)

RIBOSOME (70S)

Large Subunit (50S) --- 5S and 23S rRNAs + many proteins
Small Subunit (30S) --- 16S rRNA + many proteins

II. Eukaryotes (e.g. human)

RIBOSOME (80S)

Large Subunit (60S) --- 5S, 5.8S, and 28S rRNAs + many proteins
Small Subunit (40S) --- 18S rRNA + many proteins

1. Alkaptonuria is an autosomal recessive disorder that is associated with the production of brown sweat and dark brown urine. In a certain population, approximately 1/90,000 people are affected. What is the approximate carrier frequency for this condition?

A. 1/95

B. 1/150

C. 1/190

D. 1/300

E. 1/600

2. Autism is usually considered to be a complex trait, which affects boys 4 times more often than it affects girls. If a couple has one affected child (the proband), in which situation would the recurrence risk to a sibling be the greatest?

A. Proband is male with mild form of autism

B. Proband is male with severe form of autism

C. Proband is female with mild form of autism

D. Proband is female with severe form of autism

E. The risk would be the same regardless of proband's gender

3. Translation of the mRNA encoding the collagen polypeptide takes place at rough endoplasmic reticulum. Knowing where collagen polypeptides must ultimately end up, you would predict which of the following features for the first 20 amino acids of the just-translated collagen polypeptide?

A. The sequence will form an amphipathic helix.

B. The sequence will contain many hydrophobic residues.

C. The sequence will contain many arginine and lysine residues.

D. The sequence will contain leucine residues with requisite spacing.

E. The sequence will contain saccharide residues in place of amino acids.

4. A woman with heterozygosity for a mutation in the *dystrophin* gene (one normal and one mutant copy) is almost as severely affected with Duchenne muscular dystrophy as her maternal uncle who died at age 23 as a result of the condition. Knowing that the *dystrophin* gene is located on the X chromosome, what most likely accounts for this woman's severe symptoms?

A. She has only one X chromosome (X)

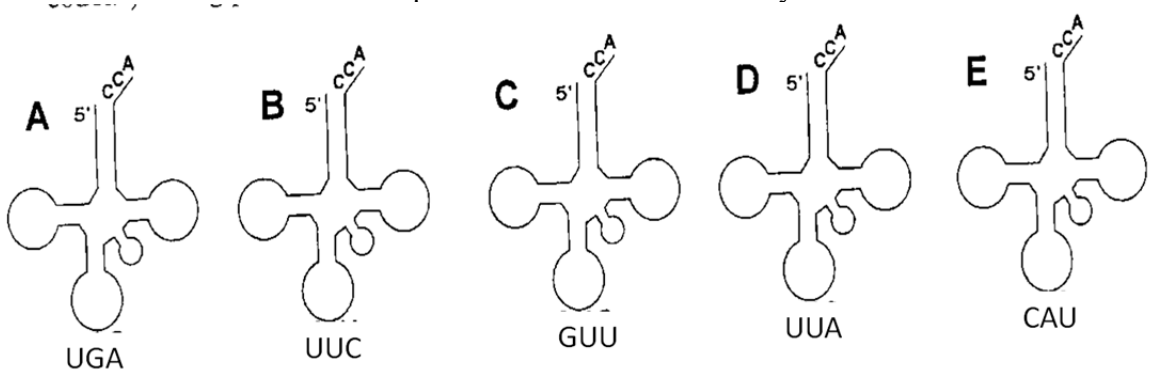
B. She has too many X chromosomes (XXX)

C. Her cells preferentially inactivated the normal X chromosome

D. She is exhibiting random, but skewed X inactivation

E. Her X chromosome is improperly imprinted

5. Which of the tRNA molecules depicted below is LEAST likely to exist in nature?



6. Upon sequencing of his genome, a healthy patient is found to express a variant of the hemoglobin beta chain in which there is a glycine to arginine amino acid change at position 83. If this Hb Muskegon variant is present in at least 1% of the population and is not associated with a change in phenotype, it is considered:

A. A genetic polymorphism

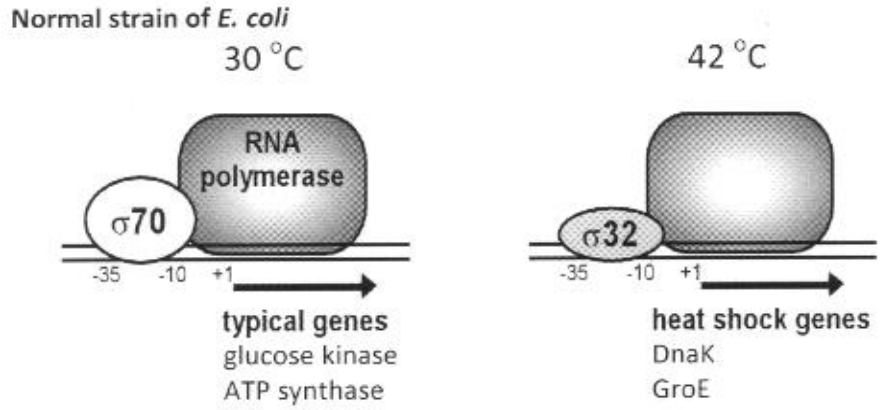
B. Locus heterogeneity

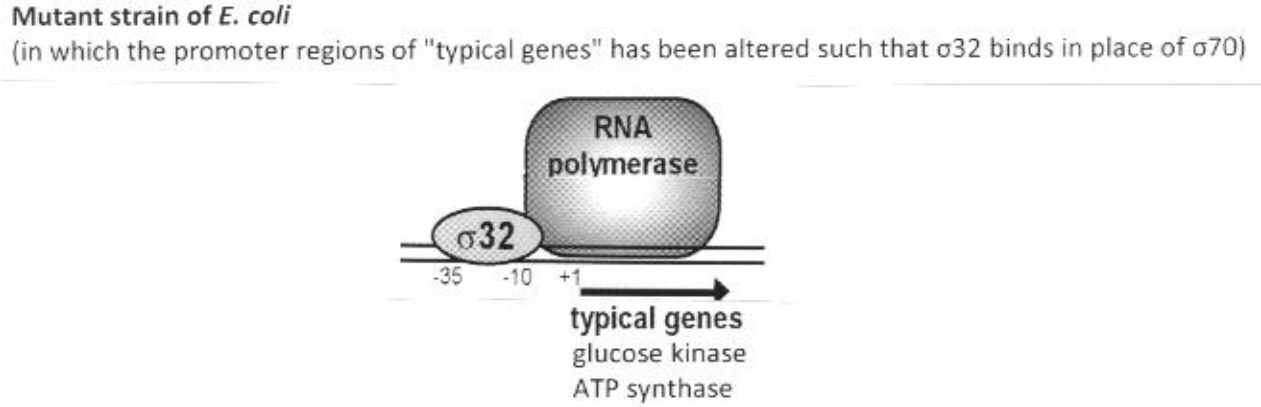
C. A silent mutation

D. A loss of function mutation

E. A phenocopy

7. In a normal strain of *E. coli*, σ70 stimulates the transcription of "typical genes" at 30 °C. The level of σ32 (an alternative sigma factor) at 30 °C is too low to allow the transcription of the "heat shock genes." When the temperature is raised to 42 °C, however, the level of σ32 rises to stimulate the transcription of the "heat shock genes." On the other hand, σ70 heat denatures and therefore, the transcription of the "typical genes" is shut off at 42 °C.





In this mutant strain of *E. coli*, the levels of the two sigma factors behave as in the normal strain: σ70 is high at 30 °C while σ32 is high at 42 °C. Which of the following correctly predicts the transcriptional status of the "typical genes" in the mutant strain of *E. coli*?

	transcription at 30 °C	**transcription at 42 °C**
A)	activated	activated
B)	activated	silent
C)	silent	silent
D)	silent	activated

8. Which of the following processes is unique to bacteria?

A. Cotranslational transport

B. Coupled transcription and translation

C. Chaperone-aided protein folding

D. Post translational protein modifications

E. Catalytic RNAs in ribonucleoprotein complexes

9. The Central Dogma of molecular biology states that one gene leads to one transcript and results in one protein product. Which of the following processes would refute this dogma?

A. Ordered splicing

B. RNA editing

C. Telomerase activity

D. Translation start site recognition

E. Polyadenylation of new transcripts

10. If a replicating cell encounters significant DNA damage during S phase, what is the most likely result?

A. It will arrest in the S phase checkpoint and undergo repair

B. It will immediately trigger apoptosis and die

C. It will utilize bypass polymerases to complete S phase

D. It will utilize telomerase to extend past the damage

E. It will undergo a reductive division

11. Angelina Jolie underwent an elective double mastectomy due to an inherited familial mutation in the gene *BRCA1* . In Miss Jolie, what DNA repair pathway would likely be defective due to a mutation in this gene?

A. Direct oxidative repair

B. Nucleotide excision repair

C. Base excision repair

D. Recombination repair

E. Nonhomologous end joining

12. What is a major purpose of wide-spread methylation of CpG dinucleotides in the genomic DNA of post-implantation fetuses?

A. Silence imprinted genes

B. Limit rearrangement of transposons

C. Reestablish totipotency of fetal cells

D. Direct differentiation of fetal cells

E. Inactivate the X chromosomes

13. Which of the following is a correct statement regarding a physical change in DNA and a possible resulting informational change?

A. A deletion is most likely to result in a missense mutation

B. Transversions are a common cause of frameshift mutations

C. An insertion will always result in a frameshift mutation

D. A transition mutation can lead to splicing errors

E. Disruptions often result in silent mutations

14. Phosphorylation of lysine residue number four on the N-terminal tail of core histone protein 2B can ultimately result in the increased expression of nearby gene clusters, such as the alpha globin genes. This is an example of what level of gene regulation?

A. Transcriptional

B. Translational

C. RNA stability

D. Post-translational

E. Chromatin packing

15. Which of the following pairs of mitotic/meiotic phases involves the splitting of centromeres?

A. Anaphase of mitosis and anaphase of meiosis II

B. Prophase of mitosis and anaphase of meiosis I

C. Anaphase of mitosis and anaphase of meiosis I

D. Telophase of mitosis and metaphase of meiosis II

E. Prophase of mitosis and metaphase of meiosis I

Use the following information to answer the next 2 questions (16-17).
A 9 month old boy presents with enlarged liver, fasting hypoglycemia, and other classic signs of Von Gierke's disease. Genetic analysis determined that both alleles of his glucose-6-phosphatase gene had the same Arg83Cys mutation. It was also discovered that his maternal great grandmother and paternal great grandfather were siblings.

16. What is the most likely class of this mutant allele and its expected inheritance pattern?

A. Gain of function mutation; dominant inheritance

B. Gain of function mutation; recessive inheritance

C. Loss of function mutation; dominant inheritance

D. Loss of function mutation; recessive inheritance

E. Loss of function mutation; codominant inheritance

17. The gene encoding glucose-6-phosphatase maps to 17q21.31. What method would be most useful to determine presence of **this mutation** in other family members of the affected boy?

A. Routine karyotyping

B. Northern blotting

C. FISH probing specific for 17q21

D. High resolution G-banding studies

E. Allele specific oligonucleotide (ASO) probing

18. Cells, when infected by a DNA virus, can transcribe both stands of the viral DNA and process it into ~21 nt single stranded fragments that can be loaded into protein complexes which target viral protein encoding transcripts for degradation. Which statement below correctly characterizes this process?

A. This siRNA silencing results in a gene knock out

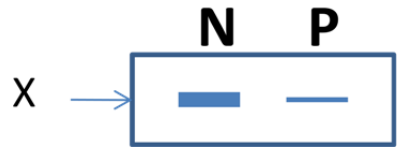
B. This siRNA silencing results in a gene knock down

C. This miRNA silencing results in a gene knock out

D. This miRNA silencing results in a gene knock down

E. This miRNA silencing results in reversible gene regulation

19. A western blot of a normal (N) and patient (P) sample is represented below. What conclusions can be drawn from these results regarding "X"?



A) The patient has less DNA for gene X relative to normal

B) The patient has less RNA for gene X relative to normal

C) The patient has less protein X relative to normal

D) The patient has more protein X relative to normal

E) The patient has more DNA for gene X relative to normal

20. The process by which a recombinant DNA (transgene) is introduced into the host is called:

A) ligation

B) recombination

C) screening

D) selection

E) transformation

21. Which of the following components is/are needed to carry out the polymerase chain reaction (PCR)?

A) A vector with a selectable marker

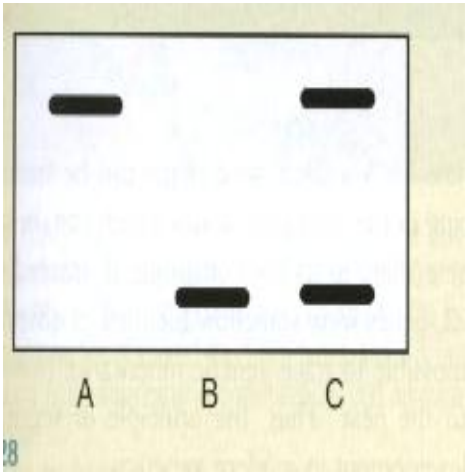
B) An origin of replication specific for the bacterium *Thermophilus aquaticus*

C) DNA ligase

D) Fluorescent or radioactive probes to detect DNA fragments of interest

E) Sense and antisense DNA primers encompassing the DNA region of interest

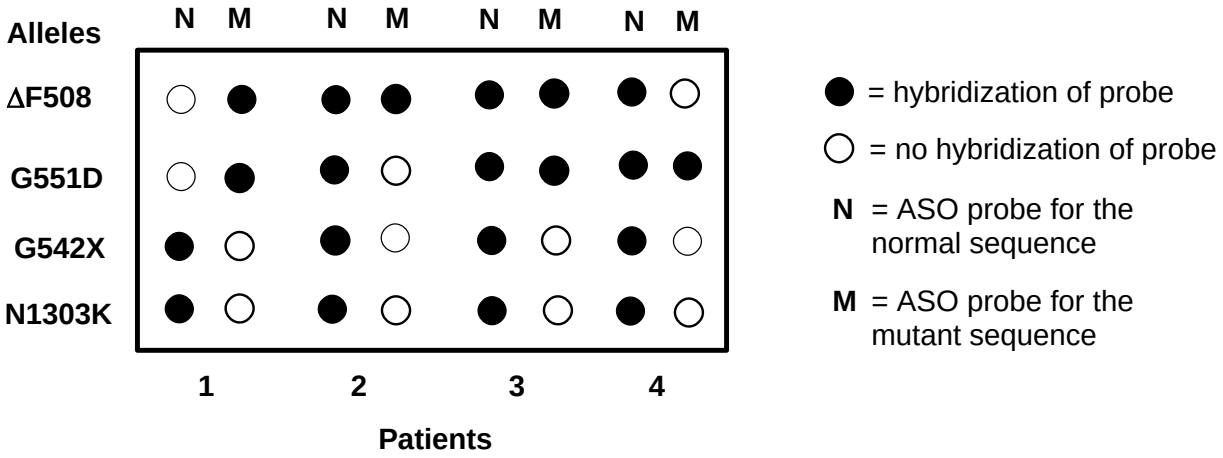
22. Alpha-1-Antitrypsin deficiency is a disease that arises when both copies of the α_1-antitrypsin gene are altered by mutations. Liver disease, chronic emphysema, and pulmonary failure can result. One of the mutations that causes α_1-antitrypsin deficiency occurs in exon 3 of the gene and destroys a recognition site for the restriction endonuclease *Bst*EII resulting in a larger DNA fragment upon digestion by that specific restriction enzyme. Using a probe that hybridizes to the region of the gene immediately upstream of that *Bst*EII restriction site, RFLP (restriction fragment length polymorphism) analysis was performed on three members of a family, producing the Southern blot result shown below. Which of the following options (A-D) best describes the genotype and disease status of individual A, individual B, and individual C?



Jorde4_F3-28_p55

Option	Individual A	Individual B	Individual C
A	Heterozygous carrier; unaffected	Heterozygous carrier, unaffected	Homozygous for mutation; affected
B	Homozygous for mutation; affected	Homozygous normal; unaffected	Heterozygous carrier; unaffected
C	Homozygous normal; unaffected	Homozygous for mutation; affected	Heterozygous carrier; unaffected
D	Homozygous normal; unaffected	Homozygous for mutation; unaffected	Heterozygous carrier; affected

23. **The figure below is an allele-specific oligonucleotide (ASO) blot of four patients tested for mutant alleles of the CF gene (coding for the cystic fibrosis transmembrane conductance regulator protein). Cystic fibrosis is an autosomal recessively inherited condition.**



The family of a 2-month old baby affected with cystic fibrosis is being tested for mutations in the CF gene. The father tests positive for being a carrier of the ΔF508 mutation and the mother tests positive for being a carrier for the G551D mutation. The 2-month-old patient carries both a deletion of the phenylalanine 508 codon (ΔF508) in one gene copy and the G551D mutation in the other gene copy. Which of the patients shown above in the ASO blot represent the ASO test outcomes for the father, the mother, and the 2-month-old (assume no recombinations)?

	Father	**Mother**	**2-month-old**
A.	patient 2	patient 4	patient 1
B.	patient 3	patient 4	patient 1
C.	patient 4	patient 2	patient 3
D.	patient 2	patient 4	patient 3
E.	patient 4	patient 2	patient 1

24. Two brothers, each with a diagnosis of some form of autosomal recessive limb-girdle muscular dystrophy (ARLGMD) marry two sisters, who also each have a diagnosis of some form of ARLGMD. Both couples each have 5 children (for a total of 10 double first cousins), none of whom go on to develop ARLGMD. The BEST explanation for this observation is:

A. Mitochondrial inheritance

B. Compound heterozygosity

C. Variable expression

D. Locus heterogeneity

E. Allelic heterogeneity

25. Your patient is a 3-week-old female in the NICU (neonatal intensive care unit). Prior to birth, she was identified to have a large ventricular septal defect (defect in the wall separating the left and right ventricles of the heart) and unilateral right kidney agenesis (congenital absence of the right kidney). Amniocentesis was performed at 19 weeks of gestation and a karyotype identified no abnormalities. After birth, respiratory distress was observed. A genetics clinic was consulted for potential determination of underlying causes for these multiple anomalies, but no obvious genetic diagnosis could be made. The BEST next test is:

A) chromosomal microarray (array CGH) analysis

B) fluorescence *in* situ hybridization (FISH) analysis specific to chromosome 7q11

C) polymerase chain reaction (PCR) using X-chromosome centromere-specific primers

D) Southern blotting for the elastin gene

E) Chromosome analysis using solid staining

26-30. **Match the following constellation of symptoms with the most likely diagnosis.**

A. Achondroplasia

B. Alport syndrome

C. Neurofibromatosis

D. Tuberous sclerosis

E. Wilson disease

26. Shagreen patches, facial angiofibromas, ash-leaf spots

27. Renal disease, hearing loss, occasional eye abnormalities

28. Liver disease, psychiatric disorders, iridal Kayser-Fleisher rings

29. Macrocephaly (large head), short limbs, “trident hand” appearance

30. Café-au-lait spots, subcutaneous nodules, axillary freckling

31. A woman planning a pregnancy is taking a new medication for migraine headaches, and wonders if this drug could have an adverse effect on her fetus, should she get pregnant while on this drug. You consult a database which lists pregnancy outcomes and fetal gestational age when the drug was taken. Which observation is most suggestive that this drug might be teratogenic? [Note: gestational age is calculated from conception]

Option	Finding in child	Gestational age
A	Growth retardation	2nd week
B	Cleft lip/palate	15th week
C	Polydactyly (extra fingers)	30th week
D	Spina bifida	4th week
E	Behavior problems	Pre-conception

32. A 6 year old boy is newly-diagnosed with autism. Additional history includes spina bifida which was repaired soon after birth. Which pregnancy history is most likely to be related to his findings?

A. Two chest X-rays in the second trimester

B. Smoked during the third trimester

C. Had a viral infection just before delivery

D. Was on valproate for seizure control throughout

E. Drank several beers the first few weeks

33. A baby is born with an abnormal skull shape, hip dislocation, and one clubbed foot. Which history would be most suggestive that the baby's anomalies are deformations?

A. Mother had a rubella infection

B. Chromosome anomaly detected on amniocentesis

C. Several family members have a p53 mutation

D. Child is one of quadruplets

E. The serum AFP level was elevated during pregnancy

The next three questions (34-36) apply to the following scenario. You have moved into a neighborhood in New Jersey which is populated by a race of aliens who had come to Earth to study the human race. They usually assume human form because they have the same genetic composition as humans, but occasionally assume their natural state for fun.

34. In their natural state, some have green spots, some have blue spots, and some have yellow spots. If spot color is a co-dominant trait, what spot color would you expect to see in offspring produced by the mating of two green-spotted individuals, if you know that green-spotted individuals are never born to parents who are either both blue-spotted or both yellow-spotted?

A. Half would have green spots and the rest yellow or blue spots

B. Half would have yellow spots and half would have blue spots

C. Half would have green spots and half would have blue spots

D. Half would have blue spots and the rest green or yellow spots

E. All would have green spots

35. You also notice that ears (at least what you think are ears) can be pointed or rounded. The rounded trait appears to be dominant over the pointed trait. If two compound heterozygotes or homozygotes for the pointed ear phenotype mate, what is the chance each offspring would have pointed ears?

A. 0%

B. 25%

C. 50%

D. 75%

E. 100%

36. Some individuals in this population develop problems involving several of their organ systems (e.g., they develop fibrosis of the splork, atrophy of the huhuma, flutz loss, and twerkopathy). What observation would be most consistent with a mitochondrial (or alien equivalent of mitochondrial) mutation as the cause?

A. Both parents need to be carriers for the condition

B. Daughters of males are all carriers

C. Males never have affected offspring

D. Males can only have affected daughters

E. Females can only have affected sons

The following three questions (37-39) apply to the following scenario. A three-generation family comes in to see you because they are interested in knowing more about the condition affecting some of the family members. Manifestations in some or all include absent spleens, intestinal malrotation, minor heart defects, extra toes, arthritis, joint dislocations, and adult-onset hearing loss and cataracts.

37. The pattern of findings can best be described as a:

A. Congenital anomaly

B. Dysplasia

C. Deformation

D. Syndrome

E. Sequence

38. What observation would be most suggestive that the mode of inheritance is autosomal dominant?

A. Males are more severely affected than females

B. The family is highly inbred

C. Only females have affected offspring

D. The phenotype is dependent on the transmitting parent

E. Males can pass the condition on to their offspring

39. Among the various manifestations, which is best characterized as being a congenital malformation?

A. Cataracts

B. Absent spleen

C. Hearing loss

D. Arthritis

E. Joint dislocations

40. A woman with Angelman syndrome caused by a microdeletion of 15q11-13 is raped in the group home in which she resides. What is (are) the expected outcome(s) of the pregnancy (assume the rapist is a caretaker, and does not have a genetic condition).

A. Angelman syndrome or unaffected

B. Prader-Willi syndrome or unaffected

C. Angelman or Prader-Willi syndrome

D. Angelman syndrome only

E. Prader-Willi syndrome only

41. An Ear, Nose and Throat (ENT) specialist sees a large family in which 5 of 8 children are homozygous for mutations in a hearing loss associated gene, CDH23. Two of the children with hearing loss are much more severely affected than are the other three; subsequently, both of the severely affected children are found to be heterozygous for a polymorphism in the ATP2B2 gene. Two of the siblings with normal hearing as well as the unaffected father also have this polymorphism in ATP2B2. The BEST explanation for the observations in this family is:

A. Reduced penetrance

B. Epistasis

C. Epigenetic modification

D. Anticipation

E. Variable expression

42. One of your patients, a 13 month old girl, is losing her ability to walk and talk. She previously had a normal head size, but now has microcephaly. In addition, she tends to keep her hands together in front of her, often appearing to wring them together. You suspect she has Rett syndrome. What is the best estimate of the chance her father's brother's newborn daughter will develop the same condition?

A. 0%

B. 1-2%

C. 25%

D. 50%

E. 100%

43. A 24 year old woman with a diagnosis of familial hypercholesterolemia relates that her mother's sister and brother, mother's father, and several maternal cousins are similarly affected. However, her mother does not have any history of elevated cholesterol levels. The BEST explanation for this observation is:

A. Locus heterogeneity

B. Clinical heterogeneity

C. Variable expression

D. Reduced penetrance

E. Mitochondrial inheritance

44. A 51 year old woman asks her primary care physician about her risk for developing autosomal dominant macular degeneration (MD), a condition associated with progressive vision loss. Her mother, mother's brother, and mother's father developed MD at 48 years, 53 years, and 46 years, respectively. The BEST information you can give her is that her risk is closest to:

A. 0%, because she is older than most of her relatives when they developed MD

B. 0%, because her mother is the affected relative

C. 50%, because MD is associated with a delayed age of onset

D. 25%, because of allelic heterogeneity in this condition

E. 100%, because she physically resembles her mother

45. A man whose two brothers and father had retinoblastoma (an autosomal dominant form of eye cancer) wants to know his chance of passing the gene to his child. The man had no history of retinoblastoma, but the penetrance is only 90%. Using Bayesian analysis, you calculate that the risk of passing the gene to his child is:

A. 9/19

B. 9/38

C. 1/2

D. 1/11

E. 1/22

46. If the penetrance of a rare, autosomal dominant form of pancreatic cancer is 60%, what is the chance that a woman with a known mutation in this gene will have an affected child?

A. 15%

B. 20%

C. 30%

D. 50%

D. 60%

47. Doug is a 58 year old man who has worked for over 20 years in a plant involved in the manufacturing of batteries. As a result of cadmium exposure, the promoter regions of several tumor suppressor genes were hypermethylated causing decreased expression of these genes. Doug is diagnosed with bladder cancer. Doug's cancer is an example of a _____ cancer that is due to a(n) _____ change. Which of the following fills the above blanks in the correct order?

A. sporadic, genetic

B. sporadic, epigenetic

C. familial, genetic

D. familial, epigenetic

E. hereditary, epigenetic

48. A pharmaceutical company has developed a new drug to treat a subtype of primary pancreatic cancers. This subtype is characterized by a genetic mutation that allows for the bypass of the G2 checkpoint in cells lacking the correct number of chromosomes. The drug targets these cells and causes apoptosis. Which the following choices best characterizes the type of gene inactivated in this subtype of pancreatic cancer?

A. Tumor suppressor

B. Proto-oncogene

C. Mismatch repair gene

D. Oncogene

E. Hereditary cancer gene

49. Which of these statements about the two-hit hypothesis is true?

A. Nonsense mutations cannot be somatic.

B. Inherited mutations predispose certain cells to cancer because there is only one functional copy of a tumor suppressor gene.

C. Somatic mutations only occur in sporadic cancers; while inherited mutations only occur in hereditary cancers.

D. An inherited mutation is only present in the cells of the tumor.

E. Two mutations in one copy of a tumor suppressor gene is sufficient to cause a cancer.

50. Beth is a 34 year old woman referred for genetic counseling because of her family history of cancer. Beth's family history is as follows: her mother was diagnosed with breast cancer at 87, her paternal aunt was diagnosed with breast cancer at the age of 41, her paternal grandmother was diagnosed with ovarian cancer at the age of 63 and died shortly after her diagnosis, and Beth's brother was diagnosed with a Wilms tumor at the age of 7. Her genetic counselor explains that the paternal family history of cancer is suggestive of a BRCA1 or BRCA2 mutation, but her mother's cancer and brother's cancer are likely unrelated to a hereditary cancer syndrome. Her genetic counselor goes on to explain that the best person for initial BRCA testing in the family is?

A. Beth's mother

B. Beth's paternal aunt

C. Beth's brother

D. Beth's father

E. Beth

51. Dr. Scarlett has found that GR1 aids in the uptake of drug X in pancreatic cancer cells. Increased uptake leads to a better treatment response. GR1 is a gene with 3 exons. Deleterious mutations in GR1 affect the response to drug X. Which of these patients with a GR1 mutation would be expected to have the **best** treatment response?

A. Patient A who has a missense mutation in exon 3 of GR1 that interrupts the binding site for drug X to enter the cell.

B. Patient B who has a silent mutation in exon 2 of GR1.

C. Patient C who has a nonsense mutation in exon 1 of GR1 that produces a truncated protein.

D. Patient D who has a missense mutation in exon 2 of GR1 that produces a protein that is degraded at a slower rate.

E. Patient E who has a large deletion that includes exons 2 and 3 of GR1.

52. Which of the following screening techniques would have the highest positive predictive value for screening for Down syndrome?
A. Ultrasound examination for soft markers (i.e. intracardiac echogenic focus)

B. Second trimester serum screening (quad screen)

C. Advanced maternal age

D. Amniocentesis with a karyotype

E. Fetal MRI

53. Confined placental mosaicism can potentially provide false results associated with which of the following tests?
A. Ultrasound

B. Chorionic villus sampling

C. Amniocentesis

D. Cell free fetal DNA analysis

E. Second trimester serum screening (quad screen)

54. A pre-symptomatic program for Huntington disease should involve which of the following:
A. Pre-test appointments with a psychologist and a genetic counselor.

B. A physical examination with a neurologist.

C. Appropriate post-test counseling.

D. Identification of a support person.

E. All of the above.

55. Mary is a healthy Caucasian woman whose mother is a carrier for cystic fibrosis. The specific mutation in Mary's mother is a deletion of exons 3 through 8. Mary is concerned about her risk of being a carrier for cystic fibrosis, so her doctor orders testing for a panel of 55 mutations in the CFTR gene (which identifies 90% of mutations in CFTR); however, this panel does not include testing for the exon 3-8 deletion. After negative testing for this panel, what is the best estimate of Mary's risk to be a carrier?

A. 3/4

B. 2/3

C. 1/2

D. 1/11

E. 0

56. Your patient, Sam, is concerned about his risk for developing stomach cancer. Sam is a 38 year old Caucasian male who has a family history of a father with stomach cancer at the age of 52. He finds two direct-to-consumer testing companies that offer genetic testing for markers associated with stomach cancer. Test one from McGenes reports that his risk for stomach cancer is increased 3 fold above the risk of an age, race, and gender matched control. Test two from Geneboree reports that his risk for stomach cancer is decreased 2 fold below the risk of an age, race, and gender matched control. This example best illustrates which of the following disadvantages of direct-to-consumer genetic testing?

A. Patient motivation

B. False advertising

C. Questionable medical benefit

D. Reliability

E. Lack of post-test genetic counseling

57. A woman has an amniocentesis and asks that a chromosomal microarray be done on the sample. The fetus is found to have a microdeletion of 15q11-13. You tell her the baby will most likely have:

A. Angelman syndrome or be normal

B. Prader-Willi syndrome or be normal

C. Angelman or Prader-Willi syndrome

D. Angelman syndrome

E. Prader-Willi syndrome

The next three questions (58-60) apply to the following situation. In the 18th century, a small group of lightly pigmented Swedish colonists moved to central Africa, where all of the population had dark skin and dark brown eyes.

58. Over the next couple of generations, some of the African population was found to have blue eyes and lighter skin color. Which factor had the greatest influence on affecting the Hardy-Weinberg equilibrium (HWE) in the African population?

A. Selective advantage

B. Gene flow

C. Non-random mating

D. New mutation

E. There is no deviation from HWE

59. A few generations later, there are fewer individuals with blue eyes and lighter skin than in the previous generations. An astute physician observes that those with blue eyes tend to be blinded by the sun (and are therefore more easily caught by lions) and those with lighter skin are more prone to skin cancer. Which factor now has the greatest influence on affecting HWE in this population?

A. Selective advantage

B. Gene flow

C. Non-random mating

D. New mutation

E. There is no deviation from HWE

60. In the mid-20th century, some individuals are found to have strikingly red hair, which appears to be inherited as an autosomal dominant trait. What factor is most likely to have influenced HWE in this population?

A. Selective advantage

B. Gene flow

C. Non-random mating

D. New mutation

E. There is no deviation from HWE

END OF EXAMINATION - Tear off this sheet and save to check your answers.

- You may write in your answer to each question on this sheet. DO NOT make any other marks on this sheet. If there are any extraneous marks on this page it will be confiscated.
- Only the answer on the scantron is the official answer. **WE CANNOT USE THE ANSWERS ON THIS TEAR OFF SHEET TO DETERMINE YOUR GRADE.**

Please remember to:

- Write in the **letter of your form** in the area titled "Period" on the exam scantron.
- **Return your examination** in the envelope provided to a proctor **before leaving the exam room**.

BMB 526 FS 2013 Exam #3

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