Exam 2 Review questions
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Note to students: These questions were formulated by myself in an attempt to recapitulate the expected spirit of exam questions. They are not pulled from previous exams and should be viewed as a general assessment of your coursework knowledge and not a conclusive or exhaustive guide to the material covered in class. That being said, I hope they help you prepare for exam 2. After the exam please let me know if you thought these questions were helpful so I can plan more effective reviews in the future.

Lecture 6

1. The karyotype of a human male with mosaic loss of the Y chromosome would contain
    a. 46 autosomes and 2 sex chromosomes
    b. 44 autosomes and 2 sex chromosomes
    c. 44 autosomes and 1 sex chromosome
    d. 44 autosomes and 1 to 2 sex chromosomes
    e. None of the above

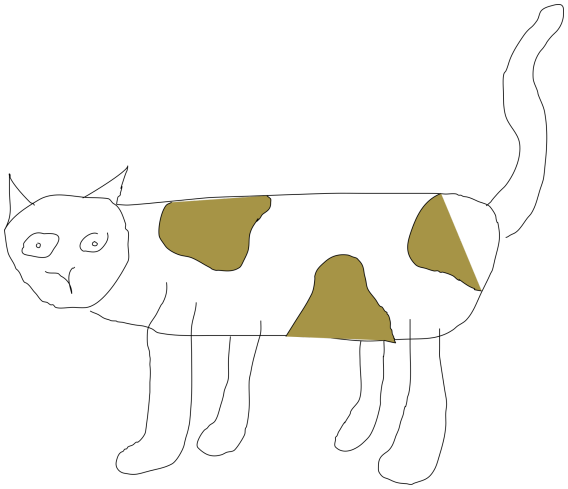
2. Name 1 X-linked disease outside of color blindness
    Rett syndrome, Fragile X, Duchenne

3. An individual with Turner syndrome would have a karyotype of
    a. 45, Y
    b. 45, X
    c. 47, XXY
    d. 22, Y
    e. 22, X

4. An individual with the karyotype 49, XXXYY would have
    a. 3 Barr bodies and 2 active Y chromosomes
    b. 2 Barr bodies and 1 active Y chromosome
    c. 2 Barr bodies and 2 active Y chromosomes
    d. No barr bodies and 2 active Y chromosomes
    e. 1 Barr body and 2 1 active Y chromosome

5. Dark pigmentation in cats is associated with the SQGE locus on the X chromosome. Cells expressing the wildtype (SQGE+) protein will produce dark pigment. What is the most likely sex of the cat shown below and why?

Female, X inactivation, 1 chromosome carries wildtype SQGE and other caries mutant



5.1. What is the most likely sex chromosome complement of the cat (incudling the version of SQGE locus carried on that chromosome) if the cat is female? If the cat is male?

Male = $X^{SQGE+}X^{SQGE-}Y$
Female = $X^{SQGE+}X^{SQGE-}$

6. An individual with the karyotype 45, Y
    a. Is likely to have delayed development
    b. Will lack RNA Pol II
    c. Will likely have higher degree of active transposon activity
    d. Is not viable
    e. Will develop normally as a male but be sterile

7. **__F__** Less than 1% of genes in the inactivated X chromosome escape inactivation.

8. **__T___**All genes that are inactivated during X chromosome inactivation will be located within the Barr Body

9. A strange transposon that carries the Xic sequence transposes into chromosome 11 of a human oocyte that is later fertilized. What will be the most likely outcome during early development of the embryo?
    a. Decrease in expression of long non-coding RNAs
    b. Global increases in transcription across all autosomes
    c. Global increase in transcription in the activated X chromosome
    d. Inactivation of both copies of chromosome 11
    e. Inactivation of 1 copy of chromosome 11

10. The Xist is considered to be the _____ of X inactivation (**Master / main switch/ regulator**

11. You create a mutant embryonic stem cell originating from a Klinefelter syndrome patient that is haploid for all autosomes and you are somehow able to keep alive. Answer the following questions
    a. What is the ratio of sex chromosomes to autosomes in this cell?
        i. **3 / 0.5 = 6:1**
    b. How many active X chromosomes would you expect in this cell?
        i. **0: Deficient in autosomal "blocking factor" due to haploid state so stoichiometry is not sufficient to shield either X chromosome from inactivation.**

12. At what location in the genome would you expect to find the highest density of Line activity?
    Xi locus

13. **__F__** During X inactivation, the two X chromosomes remain distant avoid deleterious recombination events

14. **__F__** The expression of Tsix would be expected to increase transcription at Xist

15. What is the most likely phenotype of a undifferentiated female embryonic stem cell in which is incapable of expressing Line-1 mRNA
    a. **Increase in the number of genes that escape X inactivation**
    b. Decrease in concentration of autosomal X inactivation "blocking factors"
    c. Increase in SINE element expression
    d. Increased DNA polymerase activity to accommodate lower RNA concentration
    e. Decreased frequency of transient X-X pairing

16. Deletion of the PAR region in a 46, XY cell is most likely to cause
    a. Reduction in expression from SRY locus
    b. Increase in the number of pseudogenes present on the Y chromosome
    c. Failure of sex chromosomes to pair during mieosis
    d. Decrease in expression of the Sry-Sox gene on the X chromosome
    e. Little observable effect**Lecture 7**

1. The SRY locus is also known as the _____ and is present only on the _____ chromosome.
   Testis determining factor, Y

2. Describe one method of sex determination that has evolved that differs from the human XY system.
   Birds reptiles ZW = female ZZ = male, zebrafish / turtles = environmental factors

3. An individual with the karyotype 46, XY but lacks the SRY locus will
    a. Develop normally as a male
    b. Develop normally as a female
    c. Develop female genitalia, uterus and fallopian tubes but nonfunctional gonads
    d. Develop male genitalia but have immobile sperm
    e. Not be a viable embryo

4. How do different cell types arise despite the fact that these different cell types have the same genetic material?

   Differences gene expression / regulation

5. Image selecting one cell from 2 different human individuals at random and sequencing all of the mRNA within each cell. You find the expression profiles of the two cells to be very different. These differences in transcription inferred from these datasets are most likely to be due to
    a. Selecting different cell types
    b. Differences in population specific genetic variation
    c. Differences in shared genetic variation
    d. Differences in transposon activity
    e. Differences in the location of cis regulatory elements between the genomes of the two cells

6. Intrachromosomal recombination in the Y chromosome is facilitated by
    a. High density of pseudogenes
    b. The SRY locus
    c. X degenerate sequences

d. The PAR
e. Inverted repeat sequences

7. **__F__** The human Y chromosome is more similar to chimpanzee Y chromosome than would be expected by random chance alone

Lecture 8

8. You identify a CTCF site in the human genome, the most likely genomic element to be adjacent to this site would be
    a. Catalytically associated truncation (CAT)
    b. Line 1 insertion
    c. Line 2 insertion
    d. Sine insertion
    e. Cohesion exonic sequence

9. Identify the DNA sequence most likely to result in a replication slippage event
    a. AGTGGGGAGGAAGTGAGTTT
    b. ATGCAGATGGCGATAGCAGTT
    c. GGAGGAGGAGGAGGAGGAG
    d. GGGGGGGGGGGGGGGGG
    e. AGTGGGGAAGTGGGGAAGT

D is the most repetitive sequence

10. The structural variant shown below would be most likely to be described as a

1. ATGCAGATGGCGATAGCAGTT
2. ATGCAGATG-CGATAGCAGTT

Sequence 1 is reference sequence 2 is the test sequence

    a. Indel
    b. Single nucleotide variant
    c. Copy number variant
    d. aneuploidy
    e. Not a structural variant

11. Of the choices given in question 9, which is the most commonly occurring in the human genome?

12. Which type of structural variant is most likely to affect the largest number of base pairs in the genome?

a. Ribosomal deletion variant
b. Single nucleotide variant
c. Copy number variant
d. Deletion of GGTTC
e. Indel

13. One method to measure *de novo* mutation rates would be to
    a. Sequence multiple cell types from one individual
    b. Sequence all transposable element insertions in two unrelated
    c. Sequence genomes of two fathers
    d. Sequence “quartets” of two siblings and two step parents
    e. Sequence “trios” of child plus both parents

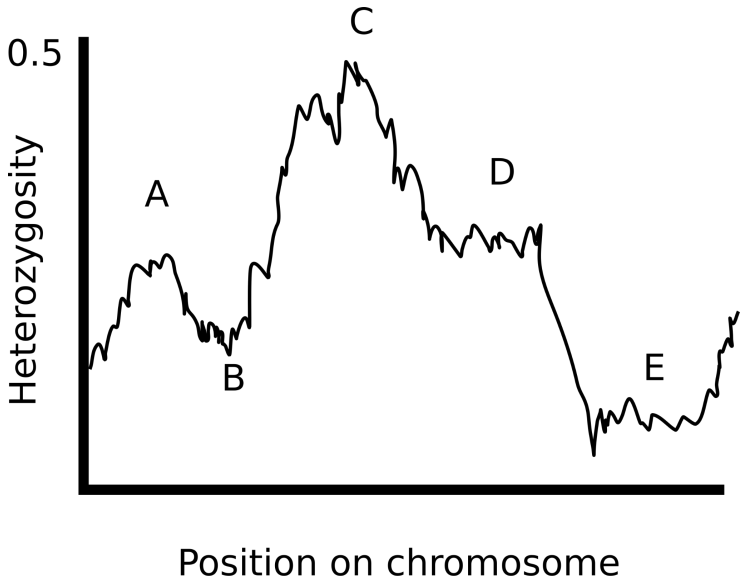
Lecture 9

1. **__F__** Most expression variation between cells is shared between continents.

2. **__F__** The chance that any given mutation will have an observable effect on phenotype is moderately high.

3. **__T__** A deleterious SNP is more likely to have originated recently than from an acient lineage.

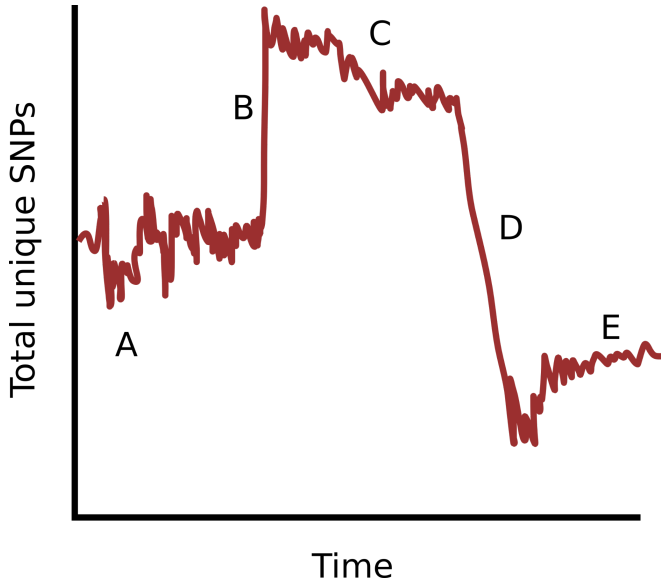
4. In the figure below identify the letter that best corresponds to the most likely locus to have been selected for during a selective sweep in the population.



   a.
   b.
   c.
   d.
   e.

5. A remote human population whose diet is entirely composed of a toxic fish has volunteered their genetic variation dataset to you. You might expect a locus that is associated with metabolism of the toxin to be
   a. Highly heterozygous
   b. Exhibit low heterozygous
   c. Not be enriched or depleted in heterozygosity

6. Select the letter which most closely corresponds to a bottleneck event in this population.



a.
b.
c.
d.
e.

8. __**T**__ The increase in total unique SNPs at point B in the previous plot may have been due to admixture with a distinct population

9. __**D**__ The decrease in total unique SNPs in the population at point D may have been due to a subset of population members separating from the original population.

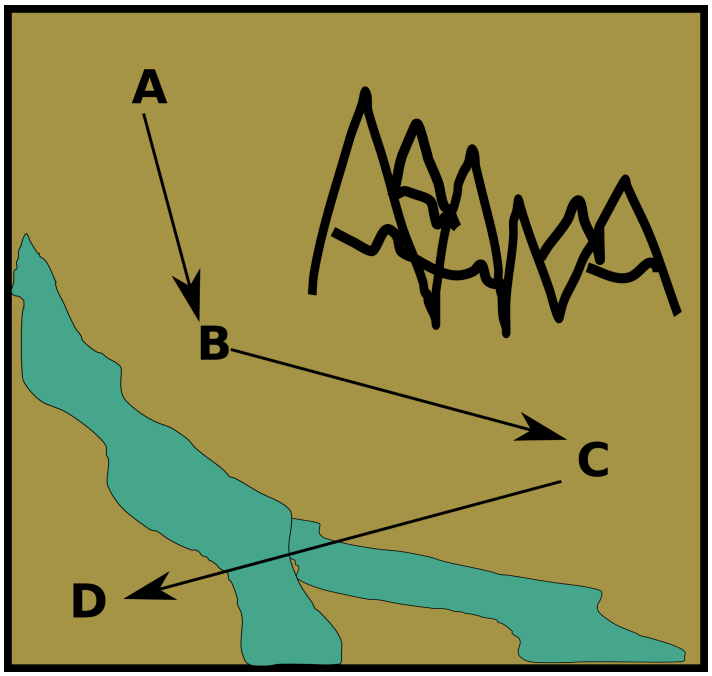
10. The phenomenon described in the previous question is known as the

a. Bottleneck effect
b. MCRA effect
c. Founder effect
d. Gooseneck effect
e. Selective sweep

8. Describe one line of evidence for the out of Africa theory of human dispersal

Mitochondrial DNA, decrease heterozygosity as distance from Africa increases

9. The image below shows a map of the migration of a population. Each letter represents an existing modern population. The later the letter in the alphabet, the earlier that population was established. Arrows represent an ancient migration event. There is no admixture between populations after a migration. Given this figure answer the questions below.



a. Which population would you expect to have the highest degree of heterozygosity? The least?
   i. **A, D**
b. If the *de novo* mutation rate in this population was zero and during each migration event exactly 1% of the population participated, what is the probability of an individual in population C having an allele that was present in 50% of the individuals in population C?
   i. **0.5*0.01*0.01 = 0.005%**
c. Provide one possible reason why migration did not occur from point B directly to point D
   i. **IDK maybe the people of B just hated water lol**