Biology 411 - Developmental Biology
Winter Quarter 2013

Midterm 1 Version B

100 Total Points
Open Book

Choose ***20*** out the 25 questions to answer (5 pts each). Only the first 20 questions that are answered will be graded. Cross out answers that you do not wish to be graded. Provide answers using full sentences, unless instructed otherwise.

1. (p. 35) Why does "acetylation" of histones promote the uncoiling of DNA? Describe the process and explain the mechanism.

Acetylation of histones neutralizes the positive charge of histones. This reduces the electrostatic interaction of histones with negatively charged DNA, resulting in an uncoiling of chromatin.

2. (p. 52) In a **female** individual with Angelman Syndrome (AS), is the Prader-Willi Syndrome (PWS) locus active? Explain why.

The Prader-Willi Syndrome locus is inactive in an individual with Angleman Syndrome. The individual lacks a portion of maternal chromosome 15, which includes both the PWS locus and AS locus. However, the intact paternal chromosome 15 has both loci. The AS locus is inactive, owing to imprinting (probably via methylation) of the gene that occurred during gametogenesis. The PWS locus is active on paternal chromosome 15.

3. (p. 99-100) If a null mutation of *lin-12* occurred in *C. elegans,* how many VPCs would form? Explain why.

Six VPCs would still form but P5.p and P7.p would most likely become central vulva cells. This is because of the lack of Lin-12 signal from the P6.p cell.

4. (pp. 152-153) Explain gamete fusion and how the egg prevents polyspermy in mice. Be specific and state protein(s)/enzyme(s) required for the sperm and egg.

Sperm docks on its side towards the PM of the egg, where the microvilli of the egg recognizes the sperm and begins fusion. Izumo (sperm) and CD9 (egg) required for gamete fusion. Slow block polyspermy only detected in mammals, controlled by release of enzymes from cortical granules. N-acetylglucosaminidase enzyme required for cleaving N-acetylglucosamines from ZP3 chains and other proteases required for modifying ZP2, both inhibiting sperm binding.

5. (p. 91) What happened to Ci in the embryonic lamb when its mother (the ewe) ate *Veratrum californicum*? What happened to transcription?

Ci remained phosphorylated in the embryonic brain cells. Ci continued to be cleaved. A portion of Ci continued to repress Hedgehog-responsive genes.

6. (pp. 80-82) Draw the resulting chick leg if wing dermis was transplanted underneath the presumptive epidermis of the embryonic foot.

Drawing: The chick's foot has wing feathers on it.

7. (pp. 54-59) What would happen if an enhancer for *myostatin* became hypo-methylated later in life, resulting in overexpression of the gene? Explain why these changes would occur.

The affected person would develop a form of muscular dystrophy (weakening of the muscles) in later life. Myostatin is needed to stop muscle growth. Too much Myostatin would result in inadequate muscle growth.

8. (p. 26) Draw a Dachshund if it had a truncated *Fgf5* gene expressed a mosaic fashion in its body.

Drawing: a normal Dachshund with patches of long hair over its body.

9. (p. 48-49) What would happen to the globin genes if Dnmt1 was inhibited during red blood cell formation?

Dnmt1 is needed to perpetuate methylation of cytosine residues on newly synthesized DNA strands, near existing sites of methylation on their template strands. Without Dnmt1 activity, methylation would not be maintained at the promoters of inactivated genes. The promoters of the epsilon-globin gene and the gamma-globin gene would be released from inactivation. Activation of a gene is possible once inactivation of the promoter is removed.

10. Trisomy 21 (Down's Syndrome) is a common birth defect in the human population. Give a plausible reason why Trisomy 20 is largely absent from the population?

Trisomy 21 results in Down's Syndrome, one of the few non-lethal trisomies. It is likely that Trisomy 20 is largely absent from the population because of embryonic lethally. Correct gene dosage is important for embryonic viability. Trisomies represent major disruptions in normal gene dosage, and many trisomies are lethal.

11. (p. 628) Two identical female twins carry one normal allele and one mutant allele for an X-linked clotting factor. One twin had severe hemophilia, whereas the other twin did not have the disease. Explain why this difference occurred in terms of epigenetics of development. Use the concept of skewing in your answer.

X inactivation in mammals occurs randomly in blastomeres during early blastocyst formation. Sometimes, a substantial fraction cells in the blastula have either the maternal X chromosome, or the paternal X chromosome, become inactivated. This situation is called skewing. In the above case, one twin had a case of skewing. The X chromosome carrying the normal allele for the blood clotting factor was inactivated in the majority of the blastomeres that gave rise to her blood-forming cells.

12. (pp. 50-52) How many Barr Bodies would be located in each somatic cells of individuals with the following genotypes? Write the number of Barr Bodies to the right of the genotype.

a. 47, XXY **1**
b. 47, YYX **0**
c. 48, XXYY **1**
d. 48, XXXY **2**
e. 46, XX **1**

13. (p. 103-105) In breast tissue, what happens when mammary gland cells became cancerous and downregulate their expression of cadherin?

The mammary gland cells reorganize the cytoskeleton, and undergo an epithelial-to-mesenchymal transition. At this point, the cells can become invasive and undergo metastasis.

14. (p. 24-25) What is the difference between analogous and homologous structures?

Analogous structures show similarities arisen from a similar function being performed, while homologous structures show similarities that were derived from a common ancestor.

15. (pp. 125-127) Is female gametogenesis complete in mammals before fertilization? Explain your answer in cellular terms.

No. The female gamete is arrested at metaphase II before fertilization. This secondary oocyte progresses through the rest of meiosis II after fertilization, to form the female pronucleus and a second polar body. (The female pronucleus combines with the male pronucleus to form the zygotic nucleus).

16. Name two epigenetic marks that are enriched on the inactivated X chromosome.

DNA methylation and histone methylation.

17. Shortened limbs could be produced by either a mutation, aneuploidy, or by teratogenesis. Explain why these different factors could produce a similar developmental abnormality.

Limb growth involves both pattern formation and morphogenesis. Each of these processes involves the interaction of numerous gene products. Mutations, aneuploidies, and teratogens can independently disrupt differential gene expression, or the function of gene products networks, leading to a failure of critical tissue patterning or tissue growth.

18. (pp. 62-64) Why are microRNAs important to embryogenesis?

miRNAs allow the activity of specific genes to be selectively suppressed, after the genes have been transcribed. miRNAs alter the activity of gene networks involved in gene differentiation.

19. (pp. 42-47) Do all transcription factors have direct contact with DNA? Explain why.

Not all transcription factors have direct contact with DNA. Some transcription factors help form bridges between the promoter and enhancer of genes, complexing with other transcription factors that actually bind to DNA.

20. (pg. 130-131) Explain the acrosome reaction in sea urchin.

Once the sperm plasma membrane contacts the jelly coat, the acrosome outer membrane fuses with the PM, leading towards acrosome exocytosis. After exocytosis, actin microfilaments push the bottom portion of the acrosome, exposing the bindin on the inner acrosomal membrane and ready for membrane fusion.

21. (pg. 51) Two types of X inactivation occur in the mouse. Briefly describe what these two types of X inactivation are and how they are different.

Imprinted X inactivation occurs in the trophoblast where the paternally derived X chromosome is preferentially inactivated. This type of X inactivation is different from X inactivation in the embryonic cells where you see random inactivation of one X chromosome.

22. (Chapter 4, p. 123) What happens to the sperm centriole after fertilization?

The single centriole provided by the sperm replicates twice after fertilization. The resulting centriole pairs become the poles of the first mitotic spindle.

23. (p. 93) If Frizzled was mutated from being an activator of Disheveled to become an inhibitor of Disheveled, what effect would this have on the overall canonical Wnt signaling pathway?

If Frizzled became an inhibitor of Disheveled, the canonical Wnt pathway would become permanently inactivated. There would be no way to inhibit GSK3. Inhibition of GSK3 is needed to allow beta-catenin to accumulate, enter the nucleus, and activate target genes. The overall canonical Wnt signaling pathway would be shut down.

24. (p. 42, Figure 2.9) What does the term "pancreas enhancer" mean?

The pancreas enhancer promotes transcription of the gene in the pancreas rudiment or pancreas organ.

25. (From discussion section and p. 39) In the film Life's Greatest Miracle, you saw an abstract, animated representation of how the *SRY* gene on the Y chromosome contributes to sexual dimorphism in the developing human fetus. In the space below, draw a rough diagram of the post-transcriptional processing of a mRNA transcript of the *SRY* gene WITHIN a cell. For your information, the SRY gene encodes a transcription factor. The gene contains introns. Using a diagram of a cell, show where nRNA and mRNA transcripts of the gene are located. Label important portions of the transcripts. More credit will be given to more complete drawings.

DIAGRAM