Solutions to 7.012 Problem Set 7

Question 1
A female geneticist has difficulty characterizing flies by their eye color because she herself is red-green colorblind (an X-linked recessive phenotype). She has one sister (colorblind) and one brother (not colorblind).

a) Sketch a pedigree that includes the geneticist, her sister, her brother, and their parents; indicate genotypes (clearly define your genotype symbols).

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  1 father Xcb/Y
  2 mother Xcb/X
  3 geneticist Xcb/Xcb
  4 sister Xcb/Xcb
  5 brother X/Y
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*bWith random mating—it’s as though there are a pool of gametes from which to choose. So imagine a pool of X chromosomes, a mixture of Xs and Xcb’s. To make a male, you pick one randomly, therefore the chance of Xcb/Y = q.

Females you pick 2 chromosomes randomly.  
\[ p^2 = \text{frequency of: } +/+ \]
\[ 2pq = \text{frequency of } cb/+; \]

b) If 8% of males in a human population are red-green colorblind, what are the frequencies of the wild-type and colorblindness alleles?
Let \( p = \text{fraction } X^+ \)
Let \( q = \text{fraction } X^{cb} \) out of total pool of X chromosomes.

To make a male, you take 1 X chromosome:

\[ \sim \text{probability } X^{cb}/Y = q \text{ and } X^+/Y = p \text{ therefore } q = 0.08 \text{ and } p = 0.92 \]

\[ c) \text{What } \% \text{ of females in this population should be red-green colorblind?} \]
\[ \sim \text{probability} = X^{cb}/X^{cb} = q^2 = (0.8)^2 = 0.64\% \]

d) What % of females in this population should be **carriers** for red-green colorblindness?
14.7% or \( X^{cb}/X = 2pq = 14.7\% \)
Question 2

a) Clearly the intracellular and extracellular environments, along with ion permeability, influence the profile of an action potential. On the diagram below, draw what you think an action potential would look in the following altered conditions. Label each action potential with the corresponding condition.

i) The external Na\(^+\) concentration is lowered.
*Lowered peak, reduced rate of rise.*

ii) The external K\(^+\) concentration is lowered.
*More negative resting membrane potential.*

iii) Scorpion toxin which slows the inactivation of Na\(^+\) channels is added.
*Prolonged repolarization*
b) Action potential wave fronts can vary even within the same neuron. You find such variation when you generated action potential graphs from recordings taken in frog motor neurons. One of the graphs represents data recorded from the axon of a neuron, while the other represents data recorded from the cell body of the same frog motor neuron. Unfortunately, you forgot to label the graphs “taken from axon” or “taken from cell body”. Given the two action potentials below, label them properly, and explain how you know your labels are correct.

Explain:
The action potential recorded from the cell body is smaller and prolonged with respect to that seen in the axon. This is because the axon is myelinated and therefore less current is leaked.
Question 3

Shown below is a diagram of a stereotypical reflex arc (particularly, the knee jerk reflex).

The sensory neurons synapse onto the interneurons and release excitatory neurotransmitters to continue the action potential through to the motor neurons.

- At which of the remaining synapses would you expect an inhibitory neurotransmitter (such as GABA) to be released? Clearly label either on the diagram or write explicitly which synapses you refer to.

  You would expect an inhibitory neurotransmitter (such as GABA) to be released at the synapse from the motor neuron serving the flexors to the flexor muscle.

  Alternatively, You would expect an inhibitory neurotransmitter (such as GABA) to be released at Synapse 7

- At which synapse would you expect an excitatory neurotransmitter (such as acetylcholine)? Clearly label either on the diagram or write explicitly which synapses you refer to.

  You would expect an excitatory neurotransmitter (such as ACh) to be released at the synapse from the motor neuron serving the quadriceps to the quadriceps muscle.
Question 4

You work in a mouse lab, studying a colony of mutant mice with a depressed phenotype: they show little interest in eating, don’t interact with other mice, and don’t play with toys that you give them.

You decide to examine the brains of some of these mice, and look at a region of the brain previously shown to be important for ‘good feelings’.

You look at synapses in this region and find that the presynaptic axon terminals of cells in the mutant brain are filled with vesicles. You compare the number of vesicles in your mutant mouse with the number in a normal mouse, and find that there are significantly more vesicles in the mutant cell.

a) What process may be affected by this mutation?

Exocytosis or Synaptic vesicle fusion

b) What neurotransmitter is likely used to transmit signals in the network of neurons you are studying?

Seratonin

c) Considering the neurotransmitter in (b), how does a mutation that affects the process in (a) explain the mutant mouse’s behavior?

If serotonin is not released to the postsynaptic cells in this region of the brain, then the postsynaptic cells are not stimulated to fire. If the postsynaptic cells are not firing, the end result of “feeling good” does not occur.

d) Could a drug that inhibited the reuptake of the neurotransmitter at the synaptic cleft relieve the mouse’s depressed symptoms? Why or why not?

No. If the neurotransmitter is never released, inhibiting reuptake will not solve the problem. Or

Yes. If a little neurotransmitter is released, inhibiting reuptake could prolong the effect of that neurotransmitter and relieve the problem.