BIOL 1114 Exam #3 (Preview) April 8, 2013

Use a #2 pencil to fill in the information on your NCS answer sheet. Put your **O-Key Account Username** in the boxes indicated **for LAST NAME** and darken the appropriate circles. **Write your Name (Last, First)** and **"Star"** or **"NoStar" in the space above the boxes containing your O-Key Account Username**. Darken the **(S)** or **(N)** in the **last column of the name circles**. Enter the number **1313** and **darken the corresponding circles** in the **first 4 columns** of the **"Student ID."** Failure to perform this correctly will incur a **-10pt handling fee**. Read all questions and answers *carefully* before choosing the **single BEST response** for each question. Feel free to ask the instructor for clarification.

		2 nd Letter								
1."	U		C		A		G		3 rd	
Letter									Letter	
U	UUU	Phenylalanine	UCU	- ·	UAU	Tyrosine	UGU	Cysteine	U	
	UUC		UCC		UAC		UGC		С	
	UUA	Leucine	UCA	Serine	UAA	STOP	UGA	STOP	A	
	UUG		UCG		UAG		UGG	Tryptophan	G	
С	CUU	J C 4 Leucine	CCU	Proline	CAU	Histidine	CGU		U	
	CUC		CCC		CAC		CGC	Arginine	С	
	CUA		CCA		CAA		CGA		A	
	CUG		CCG		CAG	Giutamine	CGG		6	
A	AUU	AUU AUC Isoleucine	ACU		AAU	Asparagine	AGU	Serine	U	
	AUC		ACC		AAC		AGC		С	
	AUA		ACA	Threonine	AAA		AGA		A	
	AUG	Methionine; START	ACG		AAG	Lysine	AGG	A rg in ine	G	
G	GUU	Valine	GCU	Alanine	GAU	Aspartate	GGU	Glycine	U	
	GUC		GCC		GAC		GGC		С	
	GUA		GCA		GAA		GGA		A	
	GUG		GCG		GAG	l Glutamate	666		6	

Cell biologists at MIT recently identified 495 proteins in the mitochondrial matrix, 31 of which had not been previously linked to the mitochondrial region. One of the newly discovered proteins, known as PPOX, is coded in a nuclear gene. PPOX is 351 amino acids long, 12% of which were glycine, the simplest of the amino acids. The PPOX gene codes for a protein responsible for the synthesis of heme, the small molecule that attaches to hemoglobin and carries oxygen in red blood cells. [Ref: Rhee, H.-W.<u>el al.</u> 2013. Proteomic mapping of mitochondria in living cells via spatially restricted enzymatic tagging. Science; Jan.]

There are about 40 distinct genetic disorders that are together referred to as Finnish Heritage Diseases because the frequencies of such disorders are much higher in Finland. One such disease is Congenital Chloride Diarrhea (CCD). It is caused by a mutation in the SLC26A3 gene that is located on chromosome 7 that hinders chloride transport. The protein encoded by the SLC26A3 gene is a transmembrane glycoprotein that transports chloride ions into the cell across the cell membrane. If the normal SLC26A3 gene codes for Glutamate as the 38th amino acid in the protein, while this gene in people with CCD has Valine instead of Glutamate at the 38th position.

A founding population of Finns settled on Åland Island that is 50 acres in the 11th century. The founding population was 50. The population grew at a rate (r) of 0.1 per decade for the first 100 years. After several decades, all the reindeer on the island were hunted to extinction, and the population size of Finns on the island for the next five decades was 98, 102, 99, 100, 101.

Historical records indicate that there was one male with CCD and another male with color blindness in the original (founding) population of Finns on Åland Island. A Health official wants to determine the frequency of CCD and color blindness in male and female Finns in the island. She researches health records and finds the following frequency data for 2013.

Sex	Frequency of CCD	Frequency of Color blindness
Male	100 in 1000	10 in 1000
Female	100 in 1000	1 in 1000

Hemophilia is a genetic disorder associated with uncontrolled bleeding due to the body's failure to manufacture a blood-clotting protein. Illustrated is the family tree of 5 generations descended from England's Queen Victoria (1820-1901) and the degree to which those descendants were affected by the **X-linked recessive gene (h)** for hemophilia. Hemophilia often (but not always) is lethal to its victims prior to reproductive age. Prince Alexie (1904-1918), son of Czar Nicholas & Princess Alexandra of Russia, was the most famous person ever afflicted with this incurable disease. None of Queen Victoria's ancestral family tree was known to have suffered from hemophilia or carried it. Prince Leopold (1853-1884), an unfortunate son of Queen Victoria, suffered from hemophilia but lived to the age of 31, married a woman (who did <u>not</u> have the **h** alleles) and produced several children.



While it is common knowledge that reptiles perform behaviors in response to temperatures or temperature changes, how they sense the temperatures is an area of investigation. A study by Seebacher and Murray (2007) identified a protein (TRPV1) that serves as an internal heat sensor and one (TRPV8) that serves as a "cool" sensor. The protein is in the membrane of sensory cells. In response to high internal temperatures the TRPV1 proteins open ion gates which (with several intervening steps) stimulates the lizard to move to cool temperatures. At low internal temperatures, TRPV8 act similarly, triggering movement toward heat. When correctly functioning, these trigger reptiles to shuttle between chambers (see illustration) with cool and

warm temperatures, which serves to regulate (maintain) their body temperatures. TRPV1 is 839 amino acids long. The lizard in the illustration below suffers from a mutation that changed the DNA codon for the 2nd amino acid in the TRPV1 protein from TTT to ATT. The mutation is found on both chromosome 17's.

Behavioral Thermoregulation Test Chamber (lizard can freely move through holes from one side to the other					
Cold Side $(10^{\circ}C)$	Hot Side $(42^{\circ}C)$				

Source of lizard: http://dnr.state.il.us/education/reptiles/lizz.htm