AP® BIOLOGY
2012 SCORING GUIDELINES

Question 3

Note: At least 1 point must be earned from each of parts (a), (b), (c), and (d) in order to earn a maximum score of 10.

Information flow in cells can be regulated by various mechanisms.

(a) **Describe** the role of THREE of the following in the regulation of protein synthesis:
- RNA splicing
- repressor proteins
- methylation
- siRNA

(*3 points maximum*)

	Description (1 point per box)
RNA splicing	• Exons spliced together. • Introns removed. • snRNPs/spliceosomes help remove introns.
Repressor proteins	• Inhibit **transcription**. • Inhibit **translation**. • Silence genes. • Inactivate gene expression.
Methylation	• DNA or histone methylation prevents transcription. • Protects against restriction enzymes.
siRNA	• Facilitates degradation of mRNA. • Inhibits translation.

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Question 3 (continued)

(b) Information flow can be altered by mutation. **Describe** THREE different types of mutations and their effect on protein synthesis.
(4 points maximum)

Type of mutation (not limited to the following)	**Description (1 point per box)**	**Effect (1 point per box)**
Silent	Nucleotide change.	No change in amino acid/protein sequence.
Missense/substitution	Nucleotide change causes new codon.	Different amino acid/protein sequence.
Nonsense/substitution	Nucleotide change causes stop codon.	Protein not formed OR truncated protein.
Frameshift (insertion/deletion)	Nucleotide insertion/deletion alters reading frame after mutation.	Changes amino acid/protein sequence OR nonfunctional protein OR no protein.
Regulatory region	Nucleotide insertion/deletion/substitution.	Alters gene expression OR alters splice site.
Translocation	Chromosome segment moves to different site.	Alters gene expression.
Nondisjunction	Chromosomes fail to separate.	
Duplication	Chromosome segment doubles.	
Deletion	Chromosome segment is removed.	
Inversion	Chromosome segment is reversed.	
Transposition	Chromosome segment moves to a different site.	

(c) **Identify** TWO environmental factors that increase the mutation rate in an organism, and **discuss** their effect on the genome of the organism.
(4 points maximum)

Environmental factor (not limited to the following) (1 point each; 2 points maximum)	**Discussion (1 point each; 2 points maximum)**
• UV light	• T-T/thymine dimers.
• Carcinogens o Cigarette smoke o Asbestos o Radon gas • Radiation o X-rays o Gamma rays/cosmic rays • Chemical mutagens o Nitrites o EtBr o Aflatoxin o Pollution	• DNA is altered/damaged (e.g., deamination, depurination, double strand breaks).
• Viruses	• Disrupt gene sequence.

Question 3 (continued)

(d) Epigenetics is the study of heritable changes in the phenotype caused by mechanisms other than changes in the DNA sequence. **Describe** ONE example of epigenetic inheritance.
(1 point maximum)

Description of an epigenetic example (1 point maximum)

Acceptable responses include, but are not limited to, the following:

- DNA or histone modifications
- Inactivated X chromosomes (Barr bodies, calico cats)
- Heterochromatin
- Tumor suppressor genes (inactivation of *p53*)
- Cellular aging
- Environmental/in utero influences
- Maternal diet
- Agouti mice
- Heavy metals
- Famine study
- Pollution
- Twin studies (e.g., identical twin variations)
- Stress-induced alterations (e.g., post-traumatic stress disorder)
- Genomic imprinting (e.g., Prader-Willi syndrome, Angelman syndrome)

3. Information flow in cells can be regulated by various mechanisms.

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(b) Information flow can be altered by mutation. **Describe** THREE different types of mutations and their effect on protein synthesis.

(c) **Identify** TWO environmental factors that increase the mutation rate in an organism, and **discuss** their effect on the genome of the organism.

(d) Epigenetics is the study of heritable changes in the phenotype caused by mechanisms other than changes in the DNA sequence. **Describe** ONE example of epigenetic inheritance.

When a segment of DNA is transcribed into RNA, the RNA can undergo splicing within the nucleus of a eukaryotic cell. A complex called a spliceosome cuts segments out of the RNA strand and joins others together. The cut segments, called introns, do not leave the nucleus and aren't expressed. The remaining segments, called exons leave the nucleus to be translated into protein. Two identical RNA molecules can be spliced in different ways and translated into or become different functional products. Repressor proteins can bind to the promoter region of an operon in DNA, preventing the attachment of RNA polymerase and/or transcription factors and preventing the expression of that gene. Repressor proteins may be always present, but capable of being activated or deactivated, making the affected operon repressible or inducible, respectively. Methylation of DNA is the attachment of methyl groups to DNA, preventing its transcription by inhibiting RNA polymerase.

Missense mutations occur when the nucleotide sequence of a gene is changed at a point in such a way that the functional product of the gene changes in composition. Nonsense mutations occur

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when a codon within a gene is replaced with a stop codon that ends translation before the rest of the gene is expressed. A frameshift mutation occurs when a number of nucleotides that is not divisible by three is deleted or inserted from/into the gene, altering the way each codon of 3 nucleotides is read. This sort of mutation changes the functional product completely in most cases.

Exposure to radiation or the intake of certain chemicals such as heavy metals increase the rate of mutation in an organism. This potentially changes the DNA permanently, affecting the products of any affected genes.

The acetylation of histone tails in the nucleosomes of a chromosome is an example of ~~mutation~~ epigenetic inheritance. The acytylation of histone proteins loosens the structure of a chromosome, increasing the rate of expression of the more exposed genes.

3B1

3. Information flow in cells can be regulated by various mechanisms.

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(b) Information flow can be altered by mutation. **Describe** THREE different types of mutations and their effect on protein synthesis.

(c) **Identify** TWO environmental factors that increase the mutation rate in an organism, and **discuss** their effect on the genome of the organism.

(d) Epigenetics is the study of heritable changes in the phenotype caused by mechanisms other than changes in the DNA sequence. **Describe** ONE example of epigenetic inheritance.

a) Repressor proteins are regulatory transcription factors that attach to the DNA helix or attach to enzymes that will attach to the DNA and prevent the DNA from unwinding and being able to be transcripted into RNA. Repressor proteins usually work of a feedback inhibition and their attachment to DNA can inhibit the synthesis of a protein. RNA splicing is a term used for the process of cutting introns. Introns are segments of DNA not needed for the production of a protein. Therefor when mRNA is created, RNA splicing "cuts out" the extra pieces of the introns.

siRNA are incredibly small proteins that help move RNA out of the nucleus, into the cytoplasm on a ribosome. There

ADDITIONAL PAGE FOR ANSWERING QUESTION 3

b) A frame shift mutation occurs when a base single base is removed/lost a frame shift occurs and the codon, read in groups of three are altered from the site of the deleted base all the way through the end of the DNA coding for a protein. Because of the new codons, a new protien or nothing at all will be produced.
when a base is added, a frame shift occurs and all the codons change from the site of the inserted base to the poly A tail of the DNA.

A substitution of a base for another base can cause a single change in the codon, and therefore the amino acid for that codon. The protein created will not be the exact protein needed because of the change in one amino acid. the only time a substitution will not cause a mutation is when the substitution will create a codon for the same amino acid.
A third cause for mution is when a segment of bases of DNA is duplicated or if a section of DNA is cut out and placed elsewhere on the chromosome. The addition of the codons (and therefore amino acids) or the switched order of codons will produce a different protein, if it produces a protein at all.

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3B3

(c) The mutation rate in an organism is affected by UV radiation and viruses.
UV radiation can ~~cause a decrease~~ degrade the bonds of a nucleotide base on the DNA, increasing the mutation rate. The genome of the organism will change with every daughter cell that the mutated cell creates. When different cells have different, alternate genomes, the organism develops a problem that may be fatal.
A virus ~~is~~ is a foreign substance to the body that inserts its DNA in our own cell and now the cell creats more viruses and virus proteins. If a virus gets in the way of a oncogen gene (genes that cancerous but turned off) it can demand the cell to divide rapidly, creating a tumor. Virus, in this case can cause cancer, like cervical cancer.

d) Epigenetic inheritance may induce.

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3C1

3. Information flow in cells can be regulated by various mechanisms.

(a) **Describe** the role of THREE of the following in the regulation of protein synthesis:

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(b) Information flow can be altered by mutation. **Describe** THREE different types of mutations and their effect on protein synthesis.

(c) **Identify** TWO environmental factors that increase the mutation rate in an organism, and **discuss** their effect on the genome of the organism.

(d) Epigenetics is the study of heritable changes in the phenotype caused by mechanisms other than changes in the DNA sequence. **Describe** ONE example of epigenetic inheritance.

a) RNA spliceosomes (complex of specialized enzymes and RNA) process mRNA before it leaves the nucleus. They remove introns (sections of RNA that stay in the nucleus) and reconnect the remaining exons (sections of RNA that exit the nucleus). By choosing which sections are introns and which are extrons, they can control the mRNA sequence that reaches the ribosomes and thus which protein sequences are made.

The addition of methyl groups to DNA causes the DNA to condense, preventing production of mRNA. Therefore, adding methyl groups to certain sequences can prevent them from being expressed.

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Repressor proteins can bind to promoters and prevent the binding of RNA transcriptase, thus stopping transcription and the expression of that gene.

b) A point mutation can change the sequence of a codon (DNA sequence for a specific amino acid) creating a faulty protein.

c) Radiation can cause a mutation in an organism, although it is usually only a part, and can lead to cancer.

Short generations spans also increase mutations, but from one generation to the next, often leading to rapid evolution. For example, in bacteria

d) AIDS can be passed through the generations from mother to child.

AP® BIOLOGY
2012 SCORING COMMENTARY

Question 3

Overview

This question asked students to describe information flow within cells and organisms — specifically, the regulation of, and the effects of mutations on, protein synthesis. Part (a) required students to describe the role of three normal cellular processes or factors in the regulation of protein synthesis, and part (b) asked for a description of three different types of mutations and the effect of the mutations on protein synthesis. In part (c) students were requested to identify environmental factors that could increase the mutation rate and to describe the effect of these mutations on the genome of an organism. Lastly, part (d) provided information about how the emerging field of epigenetics studies heritable changes in an organism's phenotype that are caused by mechanisms other than changes in the DNA sequence and then asked students to describe an example of epigenetic inheritance.

Sample: 3A
Score: 10

This response earned the maximum of 3 points in part (a). One point was earned for describing the role of RNA splicing in protein synthesis: "[S]plicosome [*sic*] cuts segments out of the RNA strand … called introns." One point was earned for describing the function of repressor proteins: "Repressor proteins can bind to the promoter[,] … preventing the expression of that gene." One point was earned for describing methylation as "the attachment of methyl groups to DNA, preventing its transcription." In part (b) the response earned the maximum of 4 points. One point was earned for describing how a missense mutation occurs "when the nucleotide sequence of a gene is changed," and 1 point was earned for describing the effect on protein synthesis: "[T]he functional product of the gene changes in composition." One point was earned for describing a nonsense mutation ("when a codon within a gene is replaced with a stop codon"), and 1 point was earned for describing its effect on protein synthesis ("ends translation"). Additional points could have been earned for the description and effect of a frameshift mutation, but the maximum for this section had already been reached. In part (c) 1 point was earned for identifying radiation as an environmental factor that increases the mutation rate, and 1 point was earned for discussing how it "potentially changes the DNA permanently, affecting the products of any affected genes." In part (d) 1 point was earned for describing an example of epigenetic inheritance: "The acetylation of histone tails in the nucleosomes … loosens the structure of a chromosome, increasing the rate of expression of the more exposed genes."

Sample: 3B
Score: 8

In part (a) 1 point was earned for describing how repressor proteins prevent DNA from being transcribed into RNA. One point was earned for describing RNA splicing as "the process of cutting introns." In part (b) the maximum of 4 points was earned. One point was earned for describing a frameshift mutation ("when a single base is removed/lost"). One point was earned for describing the effect of the frameshift: "the codon[s] … are altered from the site of the deleted base all the way through the end of the DNA coding for a protein." The response earned 1 point for describing "[a] substitution of a base for another base," and 1 point for describing the effect of a substitution ("a single change in the codon, and therefore the amino acid"). Additional points could have been earned, but the student had already reached the internal maximum of 4 points in part (b). In part (c) 1 point was earned for identifying UV radiation, and 1 point was earned for listing viruses as environmental factors that can increase the mutation rate in an organism. No points were earned in part (d).

Question 3 (continued)

Sample: 3C
Score: 6

In part (a) 1 point was earned for describing how RNA spliceosomes "remove introns." One point was earned for describing how "[t]he addition of methyl groups to DNA causes the DNA to condense, preventing production of mRNA." One point was earned for describing how "[r]epressor proteins can bind to promoters … , thus stopping transcription." The response earned 2 points in part (b): 1 point for describing how a "point mutation can change the sequence of a codon" and 1 point for describing the effect of a point mutation as a change in the "specific amino acid." In part (c) 1 point was earned for identifying radiation as an environmental factor that can cause mutations. No points were earned in part (d).