GENETICS QUESTION: 1983 L PETERSON/ECHS

State the conclusions reached by Mendel in his work on the inheritance of characteristics. Explain how each of the following deviates from these conclusions:

A. Autosomal linkage
B. Sex-linked (X-linked) inheritance
C. Polygenic (multiple-gene) inheritance

STANDARDS:
maximum = 15 points total

MENDEL (8 points maximum):
DISCRETE UNITS
2 FACTORS/TRAIT
DOMINANT/RECESSIVE

PUNNETT SQUARE:
GAMETES HAVE 1 FACTOR
EQUAL # GAMETES (EACH TYPE)
FACTORS SEGREGATE WHEN GAMETES FORM (SEGREGATION)
RANDOM DISTRIBUTION OF FACTORS (INDEPENDENT ASSORTMENT)
USE OF PROBABILITY

DEVIATIONS (12 points maximum):

AUTOSOMAL LINKAGE:	DEFINITION (1)
	EXAMPLE (1)
	EXPECTED DEVIATION (1)
SEX-LINKED:	DEFINITION (1)
	EXAMPLE (1)
	EXPECTED DEVIATION (1)
POLYGENIC:	DEFINITION (1)
	EXAMPLE (1)
	EXPECTED DEVIATION (1)

GENETICS QUESTION: 1993 L PETERSON/AP BIOLOGY

Assume that a particular genetic condition in a mammalian species causes an inability to digest starch. this disorder occurs with equal frequency in males and females. In most cases, neither parent of affected offspring has the condition.

(a) Describe the most probable pattern of inheritance for this condition. Explain your reasoning. Include in your discussion a sample cross(es) sufficient to verify your proposed pattern.

(b) Explain how mutation could cause this inability to digest starch.

STANDARDS:

Students were expected to be able to describe the most likely pattern of inheritance based on an understanding of Mendelian genetics and the specific information given to them in the question. They needed to relate that understanding to molecular genetics in their explanation of mutations as the cause for the disorder. A variety of levels of understanding to molecular genetics in their explanation of mutation as the cause for disorder. A variety of levels of understanding of the effects of mutation were accepted as students could address the mutation as affecting DNA, transcription, translation, protein structure, or protein function. Students were also expected to demonstrate their understanding of modern techniques that could detect genetic disorders.

Part A (Maximum: 4 pts)

Most Plausible Pattern:
__ autosomal (non-sex chromosome), not on X (sex-linked) or Y (holandric)
__ recessive (allele is hidden, silent or masked)

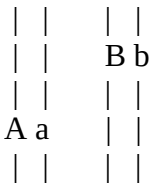
Explanation of Genetic Pattern:
__ equal frequency of condition in females and males
__ parents might not show the trait (can be heterozygous, hidden trait possible)
__ most likely a single gene is involved (only two phenotypes observed) eliminating a polygenic inheritance, etc.

Sample Cross:
__ Punnet Square, a sample cross or written explanation that substantiates their genetic pattern

Part B (Maximum: 5 pts)

Mutations:
__ gene codes for polypeptide (amylase)
__ mutations are changes in DNA or a gene
__ types of mutations (nucleotide, point or chromosomal and/or example: deletion, substitution, inversion, translocation, etc.)
__ mutations that affect transcription or RNA (RNA splicing, start signals, etc.)
__ mutations that affect translation (initiation, elongation, termination, frame shifts, etc.)
__ mutations cause protein or enzyme structural changes: missing or altered (amino acid sequence or shape, active site)
__ mutations cause protein or enzyme functional changes: (production of a nonfunctional protein)
__ mutations cause structural changes that affect the release of the active enzyme (in cell membrane or gland)

An organism is heterozygous at two genetic loci on different chromosomes.



a) Explain how these alleles are transmitted by the process of mitosis to daughter cells.
b) Explain how these alleles are distributed by the process of meiosis to gametes.
c) Explain how the behavior of these two pairs of homologous chromosomes during meiosis provides the physical basis for Mendel's two laws of inheritance.
d) Labeled diagrams that are explained in your answer may be useful – but are NOT required.

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STANDARDS: Part A (maximum of 4 points)
Transmission Via Mitosis

____ Replication- has occurred, forming 2 identical chromatids (or has occurred during S phase or interphase of cell cycle). Replication during mitosis/prophase NOT acceptable.
____ Description of chromosome alignment at metaphase. Unpaired chromosomes attached at spindle fibers (microtubules) independently of each other by metaphase.
____ Description of chromosome movement (separation of chromatids) during anaphase.

____ Description of how chromosomes move (i.e. microtubules/spindle fibers)
____ Results in 2 genetically identical daughter cells - cytokinesis.
____ Specifies that each daughter nucleus contains A, a, B, b; all alleles still present in each.
____ Elaboration-informatively labeled diagrams

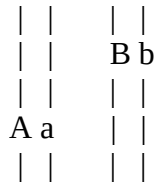
STANDARDS Part B (maximum of 4 points)
Distribution Via Meiosis to Gametes

____ Homologous chromosomes (maternal/paternal) pair (synapse) during prophase I / pairing forms tetrads.
____ Description of segregation (separation) of homologues in anaphase I.
____ No second replication between meiosis I & II / daughter cells have half the genetic material of parent cell / are haploid.
____ Description of separation of chromatids in anaphase II .
____ 2 cytokinetic events / result is 4 haploid cells or gametes produced.
____ Haploid cells / resulting cells, are not all identical.
____ Elaboration - informatively labeled diagrams

STANDARDS Part C (maximum of 4 points)
Relation of Mendel's Laws to Events in Mitosis/Meiosis

____ Correct statement of law of segregation or results of segregation ("A" separates from "a") during gamete formation. Paired elements separate randomly so each gamete receives one or other (paternal or maternal).
____ Segregation is due to separation of homologues during anaphase I.
____ Correct statement of independent assortment: during gamete formation, segregating pairs act independently of one another / AB, Ab, aB, ab.
____ Independent assortment is due to random alignment of tetrads (homologous pairs) during metaphase I (result is as likely to be AB as Ab...etc.).
____ Elaboration - Linked alleles do not show independent assortment (exception to Mendel's law). Other exceptions to Mendelian transmission : including nondisjunction etc.

An organism is heterozygous at two genetic loci on different chromosomes.



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STANDARDS: Part A (maximum of 4 points)
Transmission Via Mitosis

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____ Description of chromosome movement (separation of chromatids) during anaphase.

____ Description of how chromosomes move (i.e. microtubules/spindle fibers)

____ Results in 2 genetically identical daughter cells - cytokinesis.

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____ Elaboration-informatively labeled diagrams

STANDARDS Part B (maximum of 4 points)
Distribution Via Meiosis to Gametes

____ Homologous chromosomes (maternal/paternal) pair (synapse) during prophase I / pairing forms tetrads.

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____ No second replication between meiosis I & II / daughter cells have half the genetic material of parent cell / are haploid.

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____ 2 cytokinetic events / result is 4 haploid cells or gametes produced.

____ Haploid cells / resulting cells, are not all identical.

____ Elaboration - informatively labeled diagrams

STANDARDS Part C (maximum of 4 points)
Relation of Mendel's Laws to Events in Mitosis/Meiosis

____ Correct statement of law of segregation or results of segregation ("A" separates from "a") during gamete formation. Paired elements separate randomly so each gamete receives one or other (paternal or maternal).

____ Segregation is due to separation of homologues during anaphase I.

____ Correct statement of independent assortment: during gamete formation, segregating pairs act independently of one another / AB, Ab, aB, ab.

____ Independent assortment is due to random alignment of tetrads (homologous pairs) during metaphase I (result is as likely to be AB as Ab...etc.).

____ Elaboration - Linked alleles do not show independent assortment (exception to Mendel's law). Other exceptions to Mendelian transmission : including nondisjunction etc.

Both mitosis and meiosis are forms of cell division that produce daughter cells containing genetic information from the parent cell.

(a) **Describe** TWO events that are common to both mitosis and meiosis that ensure the resulting daughter cells inherit the appropriate number of chromosomes.

Description (1 point each; 2 points maximum)

- Spindle elements (microtubules) form/attach to chromosomes
- Chromatin condenses
- Alignment of chromosomes across center of cell prior to chromosome separation
- Separation of chromatids/centromeres to daughter cells
- G2/M checkpoint occurs in both processes
- Replication or synthesis of DNA precedes mitosis/meiosis
- Cytokinesis separates daughter cells after mitosis/meiosis

(b) The genetic composition of daughter cells produced by mitosis differs from that of the daughter cells produced by meiosis. **Describe** TWO features of the cell division processes that lead to these differences.

Feature	**Description (1 point each row; 2 points maximum)**	
	Mitosis	**Meiosis**
Number of divisions/ number of resulting cells	1 division/ 2 cells result	2 divisions/ 4 cells result
Ploidy of daughter cells	• Same as parent cell • Diploid • (2n-->2n or n-->n)	• Half of parent cell • Haploid • (4n-->2n; 2n-->n)
Chromatids separate	Occurs	Not in meiosis I/only in meiosis II
Crossing over	Does not occur	Occurs
Homologous chromosomes separate/independently assort	Does not occur	Occurs

The flow of genetic information from DNA to protein in eukaryotic cells is called the central dogma of biology.

(a) **Explain** the role of each of the following in protein synthesis in eukaryotic cells. **(5 points maximum)**

	Description (1 point each)
RNA polymerase	DNA → RNA
Spliceosomes (snRNPs)	Removes the introns and connects (splices) the exons in RNA
Codons	Codes for amino acids/signals
Ribosomes	RNA → protein or site of protein synthesis
tRNA	Transports amino acids

(b) Cells regulate both protein synthesis and protein activity. **Discuss** TWO specific mechanisms of protein regulation in eukaryotic cells. **(4 points maximum)**

Idea of the mechanism (1 point)	**Discussion (1 point)**	
Promotor	increases RNA polymerase binding	**Protein Synthesis**
Enhancer	increases transcription	
Methylation	adding methyl group inhibits transcription	
Acetylation	adding acetyl group promotes transcription	
DNA packaging	loosening/tightening chromatin promotes/inhibits transcription	
RNA processing	GTP cap or Poly-A tail	
RNA editing	removing of introns	
Alternative splicing	editing in different ways to get new/different RNA/polypeptides	
mRNA degradation	targets RNA for destruction (miRNA or siRNA)	
Protein processing	polypeptide → protein modifications (folding, chaperonins, cleavage, etc.)	
Protein degradation	proteases break down proteins	
Feedback: negative/positive	correct explanation of the identified feedback loop	**Intracellular Protein Activity**
Allosteric/noncompetitive	conformational change/binding to alternative site	
Competitive	binding to (or blocking) active site	
Environmental conditions	**intracellular** control by pH/temperature/substrate/enzyme concentration	
Phosphorylation	protein kinase/phosphorylase activating enzyme/altering 3-D shape	
Hormones	correct action for steroid or protein hormone	
Coenzymes/Cofactors	presence/absence controls reactions	

(c) The central dogma does not apply to some viruses. **Select** a specific virus or type and **explain** how it deviates from the central dogma. **(3 points maximum)**

Names a specific RNA virus or type of RNA virus (HIV, flu virus, etc.)	**(1 point)**
Deviation from the central dogma (RNA → DNA or RNA → protein or RNA → RNA)	**(1 point)**
More **detailed explanation** of the deviation from the central dogma	**(1 point)**

A difference between prokaryotes and eukaryotes is seen in the organization of their genetic material.

(a) **Discuss** the organization of the genetic material in prokaryotes and eukaryotes.

(b) **Contrast** the following activities in prokaryotes and eukaryotes

- Replication of DNA
- Transcription or translation
- Gene regulation
- Cell division

(a) 4 points maximum

(1 point for each bullet; each contrast pair must include 1 bullet from prokaryote and 1 bullet from eukaryote)

Prokaryote	Eukaryote
• No introns	• Introns
• Location: not in nucleus	• Location: nucleus (and Mito&Chloro)
• Circular	• Linear
• No histones	• Histones
• One chromosome, usually	• >one chromosome, usually
• Plasmids common	• Plasmids rare (yeast)
• Supercoiled DNA	• Chromatin DNA

(b) 8 points maximum

(each activity has a maximum of 2 points; each contrast pair must include 1 bullet from prokaryote and 1 bullet from eukaryote)

Activity	**Prokaryote**	**Eukaryote**
DNA Replication	Single origin	Multiple origin
	No telomeres	Telomeres
	Location: Cyto/Cell Memb	Location: Nucleus (and Mito & Chloro)
Transcription	No RNA Processing	RNA processing
	Location:Cyto	Location: Nucleus (and Mito & Chloro)
	Monocistronic	Polycistronic
	Initiation:sigma	Initiation: initiation factors
or	1 RNA polymerase	3 RNA polymerase
Translation	T+T coupled	T+T not coupled
	30s/40s	40s/60s
	Location: cyto	Location:also in Mito & Chloro
Gene Regulation	Operon	No operon
	+&- control	+control, primarily
	Enhancers rare	Enhancers common
	none	Methylation, acetylation, Barr bodies
Cell Division	No mitosis/Meiosis	Mitosis/Meiosis
	Rapid	Slower
	No spindles	Spindles