

GENETICS TEST I

REVIEW

- Mitosis
 - Eukaryotic cell division that yields two cells each with the same number of chromosomes as the parent cell.
- Meiosis
 - Eukaryotic cell division that produces gametes in animals and spores in plants. Meiosis converts the diploid number (2n) of chromosomes to the haploid number (n).
- Haploid
 - One set of chromosomes, 23 chromosomes in humans
- Diploid
 - Two sets of chromosomes, 46 chromosomes in humans
- Nucleotide
 - A phosphate, a sugar, and a base
 - Sugar = deoxyribose in DNA, ribose in RNA
 - Base = A (adenine), T (Thymine), G (guanine), C (cytosine) in DNA
 - U (uracil) replaces T (Thymine) in RNA
- Central Dogma
 - $DNA \rightarrow RNA \rightarrow Protein$
 - DNA \rightarrow RNA = transcription
 - RNA (mRNA) \rightarrow Protein = translation

Gregor Mendel

- Proposed the existence of particulate unit factors (genes) are passed from generation to generation
- Proposed three postulates of inheritance
 - Unit factors (alleles of genes) exist in pairs (in diploid)
 - If an organism has two different alleles (heterozygous) then one is dominant and the other is recessive. One expresses phenotype in heterozygote, and the other is hidden in heterozygote
 - The paired unit factors segregate independently during gamete formation (meiosis).
- Proposed that traits assort independently during gamete formation.
- Used true breeding strains of peas with seven characteristics, each characteristic had two contrasting traits.

- Genotype
 - The genetic makeup of an individual. The combination of alleles of an organism. (Alleles of a gene; example AA, Aa, or aa.).
- Phenotype
 - The observed characteristics.
 - Genotype + Environment \rightarrow Phenotype
- Allele
 - A variation of a gene or alternate form of a gene
- Homozygous
 - When the alleles for a trait in an individual are the same. Example AA or aa
- Heterozygous
 - When the alleles for a trait in an individual are different. Example Aa
- Hemizygous
 - Describes an individual who has only one member of a chromosome pair or chromosome segment rather than the usual two; refers in particular to X-linked genes in males who under usual circumstances have only one X chromosomeDNA

- Cytokinesis
 - The division of cytoplasm. Animal cells form cleavage furrow. Plant cells develop a cell plate.
- Karyokinesis
 - The process which chromosomes go to daughter cells by nuclear division.
- Cell cycle
 - The events that occur between completion of one cell division to the completion of the next cell division. Interphase is the beginning of cell cycle.
 - S-phase the period during which DNA is synthesized.
 - G1 period before S phase in which intensive metabolic activity, cell growth, and cell differentiation are evident.
 - G0 a period where the cell withdraws from the cell cycle. Cells in G0 stage remain viable, metabolically active, but not proliferative. (sub stage of G1)
 - G2 period after S phase in which intensive metabolic activity, cell growth, and cell differentiation are evident. Volume of cell has doubled by end of G2.
 - M Mitosis has four phases prophase, metaphase, anaphase, and telophase.
- Chromatin
 - Uncoiled DNA and protein fibers in non-dividing cells.
- Chromosomes
 - Condensed DNA & protein fibers during cell divisions. The structures in which an organisms DNA is organized.
 - A DNA molecule complexed with RNA and proteins to form a threadlike structure containing genetic information arranged in a linear sequence.
 - A structure that is visible during mitosis and meiosis.

- Kinetochores
 - A fibrous structure located with in the centromere, which is the site of microtubule attachment during cell division. (protein)
- Recombination
 - Increases genetic variation during meiosis I. Two types of recombination.
 - Crossing over exchange of DNA between homologous chromosomes.
 - Random separation (assortment/segregation) of homologous chromosomes.
 - Independent assortment orientation of homologous pairs to poles is random.
 - All possible combinations of gametes should be formed in equal frequency
 - Formula for number of combinations is 2ⁿ
- Non-disjunction
 - A cell division error in which homologous chromosomes in meiosis or sister chromatids in mitosis fail to separate and migrate to opposite poles. Responsible for defects such as monosomy and trisomy. Leads to gametes with abnormal numbers of chromosomes and can lead to abnormal offspring.
- Chromosomal Theory of Inheritance
 - The idea put forward independently by Walter Sutton and Theodore Boveri that chromosomes are the carriers of genes and the basis for the Mendelian mechanisms of segregation and independent assortment.
 - The separation of chromosomes during meiosis explains Mendel's principles of segregation and independent assortment.
- Product Law
 - In statistics, the law holding that the probability of two independent events occurring simultaneously is equal to the product of their independent probabilities.

- Sum Law
 - The sum law states that the probability of obtaining any single outcome, where that outcome can be achieved in two or more events, is equal to the sum of the individual probabilities of all such events.
- Conditional Probability
 - When one event depends on another, the likelihood of the desired outcome is the conditional probability.
- Testcross
 - A way to determine whether an individual displaying the dominant phenotype is homozygous or heterozygous for that trait. Can be used with two independent traits.
- Complete Dominance/Recessive
 - Complete Dominance is when an allele is expressed in the heterozygous.
 - Dominant allele is the allele expressed in the heterozygous.
 - Recessive when an allele is hidden in the heterozygous. An allele whose genetic expression is overridden in the heterozygous condition by a dominant allele.
- Codominance
 - Condition in which the phenotypic effects of a gene's alleles are fully and simultaneously expressed in the heterozygote.

- Multiple Alleles
 - In a population of organisms, the presence of three or more alleles of the same gene.
- Epistasis
 - Nonreciprocal interaction between nonallelic genes such that one genes influences or interferes with the expression of another gene, leading to a specific phenotype.
 - Epistasis occurs when:
 - One gene masks the effect of another gene.
 - Two gene pairs complement each other such that one dominant allele is required a each locus to express a certain phenotype.
 - Epistasis interactions often arise because two (or more) different proteins participate in a common cellular function.
- Sex linked X & Y chromosome XX female, XY male X&Y are homologous, Y chromosomes are relatively genetically inert
 - X-linked The pattern of inheritance resulting from genes located on the Xchromosome.
- Sex Influenced
 - Phenotypic expression conditioned by the sex of the individual. A heterozygote may express one phenotype in one sex and an alternate phenotype in the other sex.
- Sex limited
 - A trait that is expressed in only one sex even though the trait might not be x-linked.
 - Expression of a specific phenotype is absolutely limited to one sex. Autosomal genes expressed only in one sex.
- Y-linked
 - Mode of inheritance shown by genes located on the Y-chromosome.

Penetrance

- The percentage of individuals that show at least some degree of express of a mutant phenotype.
- Variable expressivity
 - The range of expression of a mutant phenotype..
- Complementation
 - Occurs when two strains of an organism with different homozygous recessive mutations that produce the same phenotype (for example, a change in wing structure in flies) produce offspring with the wild-type phenotype when mated or crossed. Complementation will occur only if the mutations are in different genes. In this case, each strain's genome supplies the wild-type allele to "complement" the mutated allele of the other strain's genome.
 - Complementation analysis is used to determine if two mutations causing a similar phenotype are alleles of the same gene.

Directional selection

• Phenotypes at one end of the spectrum become selected for or against, usually as a result of changes in environment.

Stabilizing selection

 Intermediate types are favored and both extreme phenotypes are selected against. This reduces population variance overtime but not the mean.

Disruptive selection

 Both phenotypic extremes are selected for and intermediates are selected against. This will result in a population with an increasing bimodal distribution for the trait.

Genetic Drift

• Random variation in allele frequency from generation to generation, most often observed in small populations.

- Nonrandom mating systems
 - Nonrandom mating systems can change genotype frequency but not allele frequency
 - Postive assortive mating similar genotypes are more likely to mate than dissimilar ones. (For example: humans are more attracted to individuals who physically resemble themselves, and are therefore more likely to be genetically similar).
 - Negative assortive mating dissimilar genotypes are more likely to mate. (Some plants have inbuilt pollen/stigma recognition systems that prevent fertilization between individuals with the same alleles at key loci.
 - Inbreeding when mating individuals are more closely related than any two individuals drawn from the population at random. Mating between relatives. Inbreeding increases the proportion of homozygotes in the population.

How many Bases are there in Dna? What are the differences between DNA and Rna?

DNA	RNA
Double Stranded	Single Stranded
Sugar is deoxyribose	Sugar is ribose
Bases are A, G, C, and T	Bases are A, G, C, and U
A pairs with T	A pairs with U
G pairs with C	G pairs with C

Examples of proteins coded for by DNA include:

Genes code for

- polypeptides (protein)
- Proteins have functions (or meaning like words)
 - Structural proteins like hair
 - Hormonal proteins like insulin and growth hormone
 - Contractile proteins like muscles
 - Enzymes (make and break covalent bonds)

Know the scientific and common names for common experimental organisms (table 1.2).

TABLE 1.2

Model Organisms Used to Study Human Diseases

Organism	Human Diseases
E. coli	Colon cancer and other cancers
S. cerevisiae	Cancer, Werner syndrome
D. melanogaster	Disorders of the nervous system, cancer
C. elegans	Diabetes
D. rerio	Cardiovascular disease
M. musculus	Lesch-Nyhan disease, cystic fibrosis,
	fragile-X syndrome, and many other
	diseases

MODEL ORGANISMS

- Model Organisms for genetic study must meet 3 criteria
 - Easy to grow
 - Short life cycle
 - Produce many offspring
- Common model organisms include
 - Escherichia coli bacterium
 - <u>Saccharomyces cerevisiae</u> yeast
 - Drosophila melanogaster common fruit fly
 - <u>Mus musculus</u> Mouse
 - <u>Caenorhabditis elegans</u> Roundworm
 - <u>Arabidopisis thaliana</u> small plant
 - <u>Danio rerio</u> Zebra Fish

What happens to the chromosomes at each stage of mitosis (interphase, prophase, metaphase, anaphase and telophase)? During which phase is DNA/duplicated/copied/doubled? During which phase does the nuclear membrane disassemble/reassemble? When does cytokinesis occur?

PHASE	Chromosome	Nuclear membrane is present/absent. Assembling or disassembling	Cytokinesis
Interphase (G2, S, G2) also G0	 Relaxed or chromatin; DNA copied during S phase of interphase 	present	No
Prophase	Chromatin condenses, chromosomes become visible.	disassembling	No
Metaphase	Chromosomes move to equatorial plane	absent	No
Anaphase	Sister chromatids separate and pulled to opposite poles of the cell.	absent	No
Telophase	Two sets of chromosome present, one set at each pole. Chromosomes begin to uncoil.	assembling	Yes

What are the differences between mitosis in plants versus animals?

	Plants (higher plants with pollen)	Animals
Centrioles (present/absent)	absent	present
Type of cytokinesis	Cell plate	Cleavage furrow

- What happens during Meiosis I?
 - Reductional Division homologous chromosomes separate.
 - Crossing over happens in prophase I
 - Random assortment alignment of each homologous chromosome is random in metaphase I
- What happens during Meiosis II?
 - Equational Division sister chromatids separate.

What are the differences between meiosis and mitosis?

	Meiosis	Mitosis
Definition	A type of cellular reproduction in which the number of chromosomes are reduced by half through the separation of homologous chromosomes, producing two haploid cells.	A process of asexual reproduction in which the cell divides in two producing a replica, with an equal number of chromosomes in each resulting diploid cell.
Function	sexual reproduction	Cellular Reproduction & general growth and repair of the body
Type of Reproduction	Sexual	Asexual
Occurs in	Humans, animals, plants, fungi	all organisms
Genetically	different	identical
Crossing Over	Yes, mixing of chromosomes can occur.	No, crossing over cannot occur.
Pairing of Homologs	Yes	No
Number of Divisions	2	1

What are the differences between meiosis and mitosis? (continued)

	Meiosis	Mitosis
Number of Divisions	2	1
Number of Daughter Cells produced	4 haploid cells	2 diploid cells
Chromosome Number	Reduced by half	Remains the same
Steps	The steps of meiosis are Interphase, Prophase I, Metaphase I, Anaphase I, Telophase I, Prophase II, Metaphase II, Anaphase II and Telophase II.	The steps of mitosis are Interphase, Prophase, Metaphase, Anaphase, Telophase and Cytokinesis
Karyokinesis	Occurs in Interphase I	Occurs in Interphase
Cytokinesis	Occurs in Telophase I & Telophase II	Occurs in Telophase
Centromeres Split	The centromeres do not separate during anaphase I, but during anaphase II	The centromeres split during Anaphase

- A cell with 46 chromosomes has how many chromosomes:
 - After mitosis in each cell?
 - 46
 - After meiosis I in each cell?
 - 23
 - After meiosis II in each cell?
 - 23
- A cell with 92 chromatids in metaphase of (mitosis or meisos) has how many chromosomes :
 - After mitosis in each ?
 - 46
 - After meiosis I in each ?
 - 23
 - After meiosis II in each ?
 - 23

- How many different gametes can be produced from the following Genotypes:
 - Vaš
 - 2
 - AaBb?
 - 4
 - AaBbCc
 - 8
 - AABbCc
 - 4
- What is a population in genetics term?
 - A local group of actually or potentially interbreeding individuals belong to the same species.
 - A group of individuals with a common set of genes that lives in the same geographic area and actively or potentially interbreed.

- What are the assumptions of the Hardy-Weinberg law:
 - Ideal population
 - No selection
 - No mutation
 - No migration into or out of the population
 - Infinitely large population
 - Random mating occurs
- The Hardy-Weinburg law makes two predictions:
 - The frequency of the alleles in the gene pool does not change over time
 - After one generation of random mating, the genotype frequencies for two alleles can be calculated as
 - $p^2 + 2pq + q^2 = 1$
 - p = the frequency of allele A
 - q = the frequency of allele a

CROSSWORD

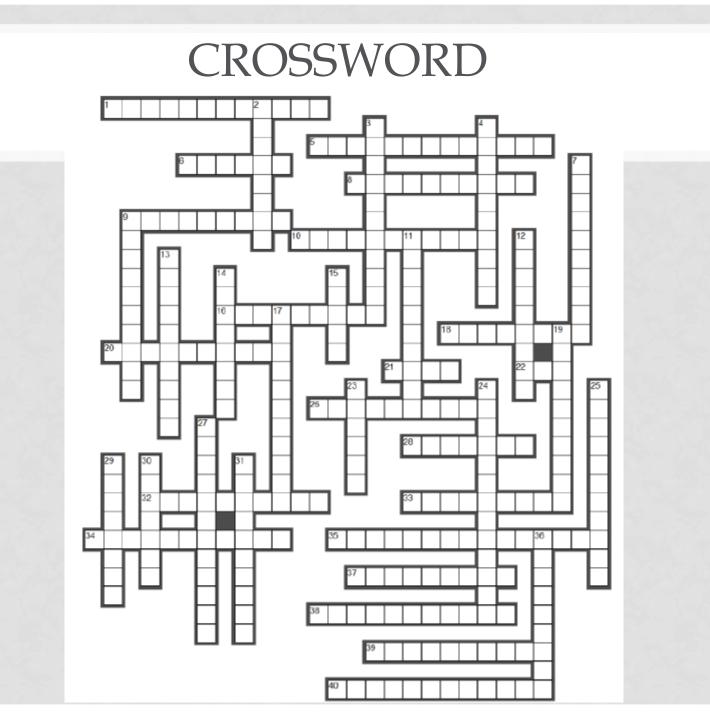
Across

- 1. An organism with two different alleles (Aa for example).
- 5. Two words no space. When a traits phenotype is modified by the sex hormones like male pattern baldness.
- Cytokinesis in animals is by a
- In the XY system where males have a gene on the X chromosome but not on the Y; then the males are not called homozygous or heterozygous but called ____.
- 9. The observable trait of an organism.
- 10. If beak size got larger and larger over many generations then this would be an example of _____ selection
- 16. During this phase of the mitosis the chromosomes are at the equator
- During this phase of mitosis the chromosomes are condensing.
- When the heterozygote expresses both phenotypes.
- The end result of meiosis I is two cells with ____ the number of chromosomes as the original cell
- In mammals XX are females and XY are males. X and Y are called the ____ chromosomes.
- Adds repeated units of DNA to the ends of chromosomes.
- 28. Having one set of chromosomes
- 32. The percentage of individuals with a mutant genotype that express the mutant phenotype to some degree.
- A trait coded for by a gene on the X chromosome is ____ (two words no spaces).
- When chromatids separate they are now called ____.
- 35. More than two alleles (two words no spaces).
- 37. Relaxed DNA found in interphase nucleus of a eukaryotic cell
- If beak size stayed the same size over many generations as larger and smaller beak birds died, then this
 would be an example of _____ selection
- 39. An organism with two of the same allele (for example AA or aa).
- Division of nuclei only (term does not include division of cytoplasm).

CROSSWORD

Down

- 2. Combination of alleles of genes is called _____
- This is the where spindle fibers attach to the bud of protein on the centromere of a chromosome.
- When the heterozygote expresses a phenotype in-between both homozygotes, the trait is _____ dominant.
- If a population of birds with average beak size formed two populations of birds with larger and another population with smaller beak size then this would be an example of ____ selection.
- 9. A mutation in one gene has multiple phenotypes.
- 11. During S portion of this phase of the cell cycle DNA is being replicated.
- 12. During this phase of mitosis the chromosomes are relaxing
- 13. Adjective for when two chromosomes are of the same size and same shape and pair up during meiosis.
- 14. When the heterozygote has the same phenotype as a homozygote, then that trait is said to be completely
- 15. Cytokinesis in plants is by forming a cell
- 17. Plants have an _____ of generations (between the haploid gametophyte and diploid sporophyte).
- 19. Two words no spaces. Term for when only one sex exhibits a trait like cows give milk but not bulls.
- A variation of a gene is called an ____.
- This is the word describing the new combinations of chromosomes resulting from the process of meiosis I
- Cell separation or division of the cytoplasm after mitosis or meiosis is called _____.
- When all individuals have mutant phenotype but vary in range of expression of the phenotype (some more severe than others).
- 29. During this phase of the cell cycle the chromatids separate and are now called chromosomes.
- 30. having two sets of chromosomes (one set from each parent)
- 31. This separates in mitosis and meiosis II (plural form of word).
- 36. For example, when one gene masked the effect of another.



- An organism with two different alleles (Aa for example.
 - Heterozygous
- When a traits phenotype is modified by the sex hormones like male pattern baldness.
 - Sex Influenced
- Cytokinesis in animals is by a _____
 - Furrow
- In the XY system where males have a gene on the X chromosome but not on the Y; then the males are not called homozygous or heterozygous but called

Hemizygous

- The observable trait of an organism.
 - Phenotype
- If beak size got larger and larger over many generations then this would be an example of _ selection.
 - Directional
- During this phase of the mitosis the chromosomes are at the equator
 - Metaphase
- During this phase of mitosis the chromosomes are condensing.
 - Prophase

- When the heterozygote expresses both phenotypes.
 - Codominant
- The end result of meiosis I is two cells with _____ the number of chromosomes as the original cell
 Half
- In mammals XX are females and XY are males. X and Y are called the ____ chromosomes.

• Sex

- Adds repeated units of DNA to the ends of chromosomes.
 - Telomerase
- Having one set of chromosomes
 - Haploid

- The percentage of individuals with a mutant genotype that express the mutant phenotype to some degree.
 - Penetrance
- A trait coded for by a gene on the X chromosome is _____
 - Sex Linked
- When chromatids separate they are now called
 - Chromosomes
- More than two alleles
 - Multiple Alleles
- Relaxed DNA found in interphase nucleus of a eukaryotic cell
 - Chromatin

- If beak size stayed the same size over many generations as larger and smaller beak birds died, then this would be an example of ______ selection
 - Stabilizing
- An organism with two of the same allele (for example AA or aa).
 - Homozygous
- Division of nuclei only
 - Karyokinesis
- Combination of alleles of genes is called____
 - Genotype
- This is where spindle fibers attach to the bud of protein on the centromere of a chromosome.
 - Kinetochore

- When the heterozygote expresses a phenotype inbetween both homozygotes, the trait is _____ dominant
 - Incomplete
- If a population of birds with average beak size formed two population of birds with larger and another population with smaller beak size this would be an example of ______ selection
 - Disruptive
- A mutation in one gene has multiple phenotypes.
 - Pleiotropy
- During S portion of this phase of cell cycle DNA is being replicated
 - Interphase
- During this phase of mitosis the chromosomes are relaxing
 - Telophase

- Adjective for when two chromosomes are of the same size and same shape and pair up during meiosis.
 - Homologous
- When the heterozygote has the same phenotype as a homozygote, then that trait is said to be completely ____
 - Dominant
- Plants have an _____ of generations (between the haploid gametophyte and diploid sporophyte).
 - Alternation
- Term for when only one sex exhibits a trait like cows give milk but not bulls.
 - Sex Limited
- A variation of a gene is called an _____
 - Allele

- This is the word describing the new combinations of chromosomes resulting from the process of meiosis I.
 - Recombination
- Cell separation or division of the cytoplasm after mitosis or meiosis is called ______
 - Cytokinesis
- When all individuals have mutant phenotype but vary in range of expression of the phenotype (some more severe than others).
 - Expressivity
- During this phase of cell cycle the chromatids separate and are now called chromosomes.
 - Anaphase
- Having two sets of chromosomes (one set from each parent)
 - Diploid

- This separates in mitosis and meiosis II.
 - Chromatids
- For example, when one gene masked the effect of another.
 - Epistasis



Questions



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