

VOCABULARY REVIEW

Gene = a unit of heredity that is transferred from parent to offspring and is held to determine some characteristic of the offspring (factors that control a trait)

Heredity = the transmission of characteristics from parent to offspring

Genetics = the scientific study of heredity

Trait = a characteristic that an organism can pass on its offspring through its genes

Purebred = an organism that is the offspring of many generations that have the same trait

Alleles = the different forms of a gene; each parent donates one allele for every gene

Dunnett square = a chart that shows all the possible combinations of alleles that can result from a genetic cross between 2 parents

Fertilization = the process where an egg and sperm cell join to form a new organism

Probability = the number that describes how likely it is that an event will occur
↳ # of times an event is expected to happen
of times an event could happen (total)

Ratio = The relationship between 2 numbers being compared to one another

Heterozygous/Hybrid = an organism that has 2 different alleles for a trait

Dominant Allele = an allele whose trait shows up in the organism when the allele is present

Recessive Allele = an allele that is hidden whenever the dominant allele is present

Homozygous = an organism that has 2 identical alleles for a trait

Genotype = an organism's genetic makeup or allele combination

Phenotype = an organism's physical appearance; visible traits

Genome = the full set of genetic information that an organism carries in its DNA

Karyotype = the complete diploid set of chromosomes grouped together in pairs arranged in order of decreasing size

GREGOR MENDEL

Gregor Mendel was the father of genetics

He studied pea plants and noticed they all had different traits

- Law of Segregation:**
- 1) traits are handed down through "hereditary factors" in the sperm and egg
 - 2) Because offspring obtain hereditary factors from both parents, each plant must contain 2 factors for every trait (alleles)
 - 3) The factors in a pair segregate during the formation of sex cells, and each sperm or egg receives only one member of the pair

- Principles of Heredity:**
- 1) The inheritance of biological characters is determined by individual units called genes, which are passed from parents to offspring
 - 2) When 2 or more forms (alleles) of the gene for a single trait exist, some alleles may be dominant and others may be recessive
 - 3) In most sexually reproducing organisms, each adult has 2 copies of each gene - one from each parent
 - 4) The alleles for different genes segregate independently of each other when sex cells are formed

Even though Mendel worked with plants, many biologists had tested all of Mendel's principles and learned that they applied to other organisms as well

Ex. Thomas Hunt Morgan used fruit flies (*Drosophila melanogaster*) and tested Mendel's principles

- Independent Assortment:** Genes for different traits can segregate independently during the formation of gametes and do not influence each other's inheritance
- Independent assortment is the reason for many genetic variations in organisms - even when they have the same parents
- It is chromosomes that assort independently not individual genes

BLOOD TYPES IN HUMANS

Human blood comes in a variety of genetically determined blood groups

A number of genes are responsible for human blood groups

The best known are the ABO blood groups and the Rh blood groups

3 alleles possible (A, B, and O) (letters stand for specific antigen) but each person only has 2 alleles

A and B are codominant and O is recessive $A = I^A$ $B = I^B$ $O = i$

6 combinations (genotypes) $I^A I^A$ or $I^A i = A$ $I^B I^B$ or $I^B i = B$ $I^A I^B = AB$ $i = O$ 4 phenotypes $\Rightarrow A, B, AB, \text{ and } O$

Rh factor: positive ($Rh+$) is completely dominant over negative ($Rh-$)

$Rh+$ or + = dominant $Rh-$ or - = recessive

Rh positive blood = ++ or - Rh negative blood = --

DUNNETT SQUARES

One factor cross

Step 1: Start with the parents \rightarrow Bb and Bb

Step 2: figure out the gametes \Rightarrow Bb \searrow B
b \swarrow b-Bb

Two factor cross

TtGg and TtGg

| | | | | |
|------|---|---|---|---|
| TtGg | $\frac{1}{4}$ T ⁺ G ⁺ | $\frac{1}{4}$ T ⁺ G ⁻ | $\frac{1}{4}$ T ⁻ G ⁺ | $\frac{1}{4}$ T ⁻ G ⁻ |
| | T ⁺ G ⁺ | T ⁺ G ⁻ | T ⁻ G ⁺ | T ⁻ G ⁻ |

Step 3: line them up \rightarrow

| | |
|---|---|
| B | b |
| B | |
| b | |

| | T ⁺ | G ⁺ | T ⁻ | G ⁻ |
|----------------|----------------|----------------|----------------|----------------|
| T ⁺ | | | | |
| G ⁺ | | | | |
| T ⁻ | | | | |
| G ⁻ | | | | |

Step 4: write out the new genotypes \rightarrow

| | |
|--------------|--------------|
| B \times b | B \times B |
| B \times b | b \times b |

| | T ⁺ | G ⁺ | T ⁻ | G ⁻ |
|----------------|----------------|----------------|----------------|----------------|
| T ⁺ | | | | |
| G ⁺ | | | | |
| T ⁻ | | | | |
| G ⁻ | | | | |

Step 5: figure out the results \rightarrow

| | |
|----|----|
| B | b |
| BB | Bb |
| Bb | bb |

3/4 = large beaks

1/4 = small beaks

1/2 = homozygous

1/2 = heterozygous

| | T ⁺ | G ⁺ | T ⁻ | G ⁻ |
|----------------|----------------|----------------|----------------|----------------|
| T ⁺ | | | | |
| G ⁺ | | | | |
| T ⁻ | | | | |
| G ⁻ | | | | |

tall yellow = 9/16

short yellow = 3/16

tall green = 3/16

short green = 1/16

* If both parents are heterozygous for both traits, (dihybrid \Rightarrow 2 factors cross) the ratio will always be 9:3:3:1

OTHER PATTERNS OF INHERITANCE

Incomplete Some alleles are neither dominant or recessive

dominance: The heterozygous phenotype lies in between the 2 homozygous phenotypes (blend)

Ex. PP = red WW = white RW = pink (mix of red + white)

codominance: the phenotypes produced by both alleles are clearly expressed

Ex. blood type: AB blood BB = black bb = white Bb = black with white stripes

Many genes exist in several different forms and are therefore said to have multiple alleles

Ex. blood type (3 alleles possible \Rightarrow A, B, and O) but each person has only 2 of these alleles

Many traits are produced by the interaction of several genes

Traits controlled by 2 or more genes are called polygenic traits

Ex. variety of human skin color \Rightarrow more than 4 genes control skin color

Genes provide a plan for development, but how that plan unfolds also depends on the environment

HUMANS AND GENETICS

Humans have 46 chromosomes $2N=46$

Two of the 46 chromosomes are known as sex chromosomes because they determine an individual's sex

Female = XX (2 copies of X chromosome) Male = XY (one X chromosome + one Y chromosome)

The remaining 44 chromosomes are known as autosomal chromosomes, or autosome

Sex determination: All human eggs carry one X chromosome (22 autosomes, and one X)

$\frac{1}{2}$ of all sperm carry an X chromosome (22 autosomes and one X) and $\frac{1}{2}$ carry a Y chromosome (22 autosomes, and one Y)

Always 50% of girl offspring and 50% of boy offspring

| | | | |
|---|----|----|----------|
| x | x | x | 50% girl |
| y | xy | xy | 50% boy |

Sex-linked gene = gene located on a sex chromosome

X-Chromosome Mary Lyon discovered that in female cells, one X chromosome is randomly

Inactivation = switched off, forming a dense region in the nucleus known as a Barr Body

Barr bodies are not found in males because their single X chromosome is still active.

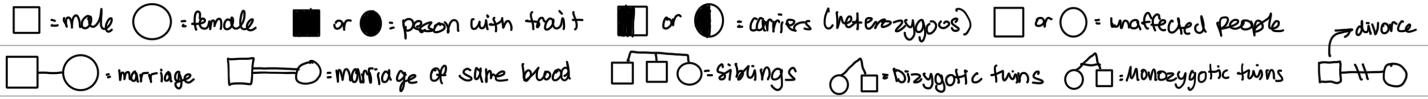
PEDIGREE CHARTS

Shows the relationships within a family

Diagram shows how a trait is inherited over several generations

Chart helps genetic counselors analyze pedigree charts to infer the genotypes of family members

Makes it possible to determine the nature of genes and alleles associated with inherited human traits



Dizygotic twins = non-identical twins (two sperms fertilizing 2 eggs) Share 50% of genes (like any sibling)

Monzygotic twins = identical twins (one egg fertilized by one sperm, but splits into 2) Share 100% genes (alone)

Autosomal Appears in both sexes with equal frequency

Recessive = Trait tends to skip generations

Affected offspring are usually born to unaffected parents

Appears more frequently among the children of related marriage

Ex. albinism, cystic fibrosis, Tay-Sachs

Autosomal Appears in both sexes with equal frequency

Dominant = Both sexes transmit the trait to their offspring

Does not skip generations

Affected offspring must have an affected parent unless they have a mutation

Unaffected parents do not transmit the trait

Ex. Achondroplasia, Huntington's Disease, Hypercholesterolemia

X-linked both males and females are affected; often more females than males are affected

Dominant = Does not skip generations

Affected sons must have an affected mother

Affected daughter must either have an affected mother or an affected father

Affected fathers will pass the trait on to all daughters

Affected mothers (heterozygous) will pass trait to $\frac{1}{2}$ sons and $\frac{1}{2}$ daughters

Ex. Fragile X syndrome, Hypophosphatemic Rickets

X-linked : more males than females are affected

Recessive Affected sons are usually born to unaffected mothers thus the trait skips generation

$\frac{1}{2}$ of carrier mothers' sons are affected

Never is passed from father to son

All daughters of affected fathers are carriers

Ex. Color blindness, Hemophilia, Duchenne Muscular Dystrophy

Sex-linked disorders are more common in males than females because for a recessive allele to be expressed in females there must be 2 copies of the allele, one on each of the 2 X chromosomes. Males have just one X chromosome. Thus all X-linked alleles are expressed in males even if they are recessive.

Hemizygous = having or characterized by one or more genes that have no allelic counterparts

Ex. X chromosome being paired with Y or genetic deficiency

The X chromosome contains genes that are vital for the survival/development of the embryo

GENETIC DISORDERS

Changes in a gene's DNA sequence can change proteins by altering their amino acid sequences which may directly affect one's phenotype.

There is a molecular basis for a genetic disorder

lethal recessive having 2 copies of an essential gene may be deadly

alleles = Ex. cystic fibrosis

If 2 copies of an autosomal chromosome fail to separate during meiosis, nondisjunction, an individual may be born with 3 copies of a chromosome

Down syndrome = 3 copies of chromosome # 21

Turner's syndrome = females only have one X chromosome (can't reproduce)

Klinefelter's syndrome = extra X chromosome in males (interferes in meiosis, preventing reproduction)

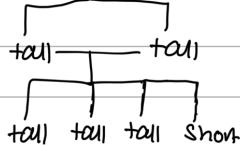
OTHER INFO FROM TEXTBOOK

- Mendel cross-pollinated peas for his experiment (peas = true-breeding = asexual reproduction)

↳ cut away male pollen and dusted onto female part

Mendel's experiments

results: tall | tall



He chose pea plants because they are small and easy to grow

And they can reproduce quickly

from molecule to phenotype: 1) gene's DNA sequence 2) The amino acid sequence changes 3) A change in that alters a protein changes phenotype results

more than 1200 genes are found on the X chromosome
but only 140 genes on the Y chromosome

Trisomy = genetic disorder when a person has 3 copies of a chromosome instead of 2

Sickle Cell Disease = caused by defective allele for beta-globin, one of the polypeptides in hemoglobin

makes hemoglobin molecules to stick together when oxygen level decreases
tends to get stuck in capillaries and can damage body

Cystic Fibrosis = caused by deletion of 3 bases in CFTR gene

CFTR allows chloride ions to pass across cell membranes

The loss of the bases removes phenylalanine amino acid and causes the protein to fold improperly
This prevents the transport of chloride ions

Huntington's Disease = caused by a dominant allele for a protein found in brain cells

The allele for this disease codes CAG (amino acid glutamine) repeatedly over 40 times
mental deterioration and uncontrollable movements

Hemophilia = caused by an absent blood-clotting protein

stable clots do not form quickly over wounds, causing extensive bleeding

sex-linked disorder (on X chromosome)