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Chapter 11 Genetics: The Work of Mendel page 308

All organisms have a set of Chacteristics inherited from their parents

Heredity: the delivery of characteristics from parents to offspring

Genetics: the study of heredity

Gregor Mendel (considered to be the Father of Genetics)

- An Austrian Monk
- Worked in the Monastery gardens
  - Studied garden pea
  - Thus changed Biology forever

~Studying of pea plants~

True-Breed: demonstrates 1 form of a trait

Self-Pollination: flower that pollinates (fertilizes) itself

- Thus the traits of this offspring should be the same
- **Trait=** a specific chacteristic of an individual
- However WHY do some differ?

Cross-Pollination: 1 flower fertilizes another

- This creates a hybrid
  - The offspring of a cross between parents with different traits

~Generation of Genetic Crosses~

Parental Generation or Pi

- These are the orginal parents
- These are pure for their trait
- How were these crossed: 2 pure breeds crossed (1 dom & 1 rec)

First Filial or Fi

- These are the offspring of the Pi generation
- All of these show the dominant trait
  - Why do they only show this trait?
  - **MENDEL"S CONCLUSION:** an individual chacteristics are determined by factors that are passed from 1 parental generation to the next
  - **These factors are called genes:** factors passed from parent to offspring
  - He concluded that there must be different contrasting forms of a gene
    - The different forms of a gene is called an Allele
      - There are 2 alleles for every trait that you have

This also led to CONCLUSION #2: Principle of Dominance

- Some alleles are considered to be dominant and some are considered to be recessive

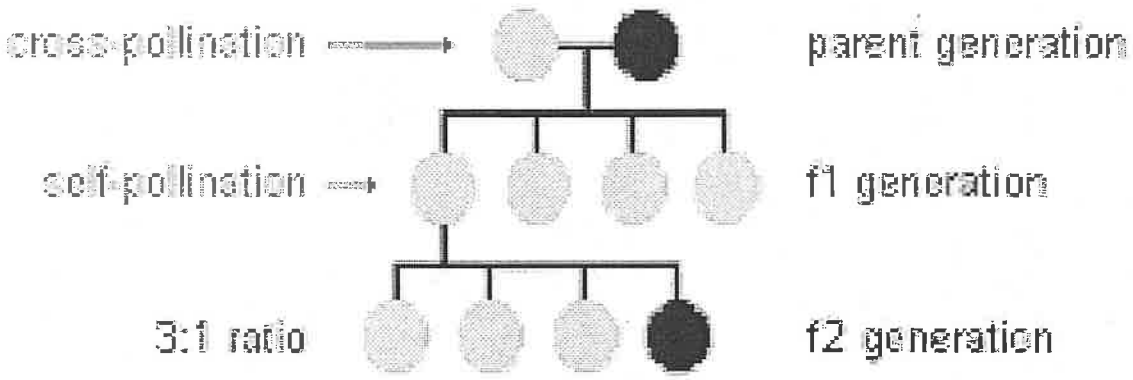
- **DOMINANT:** form of a trait that appears
  - Must be written with a capital letter
  - Only needs to have 1 out of the 2 alleles to be considered dominant
    - Examples: T, F, R, B
- **RECESSIVE:** form of trait masked by dominant
  - Must be written with a lowercase letter
  - Must have 2 recessive alleles to be considered recessive
  - How can we prove that there are alternate forms of a gene??

- The First Filial Generation was then self-fertilized

Second Filial or the F_2

- These are the offspring of the F_1 generation
- All of a sudden he found the recessive trait showed up
  - There was 3 (75%) dominant
  - There was 1 (25%) recessive
  - How can this be?
  - **MENDEL'S CONCLUSION = LAW OF SEGREATION**
    - During gamete formation the alleles for each gene segregate from each other, so that each gamete carries only 1 for each gene
    - Thus F_2 generation must have 1 of each allele but the dominant is seen
- All of these are in a 3:1 ratio 3 dominant to 1 recessive

Order of generations P_1 → F_1 → F_2



~Applying Mendel's Principles~

Probability: The likelihood that a particular event will occur

Calculations: $\frac{\text{Number of 1 kind of possible outcome}}{\text{Total number of all possible outcomes}}$

Practice:

Homozygous: organisms that have *2 identical alleles* for a particular gene

2 types

1) Homozygous Dominant: 2 dominant alleles
   a. Examples RR TT BB
2) Homozygous Recessive: 2 recessive alleles
   a. Examples: rr tt bb

Heterozygous: organisms that have *2 different alleles* for the same gene

Thus there must be 1 dominant and 1 recessive allele

Examples: Rr Tt Bb

Phenotype: the *physical* characteristics of an organism

This is what is physical seen or observed

Genotype: the *genetic* make-up of an organism

This is the allele combination

Punnett Square: this diagram uses mathematical probability to help predict the genotype and phenotype combinations of a genetic cross

Monohybrid Cross: when 1 particular trait is crossed between a male and female to identify all possible outcomes

- Has 3 possible genotypes
- Has 2 possible phenotypes

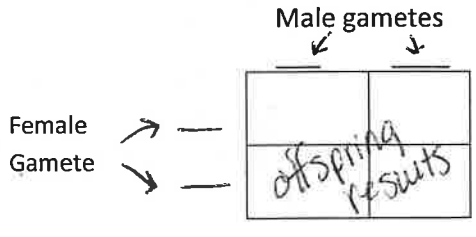
Dihybrid Cross: when 2 traits are simultaneously crossed between a male and female to identify all possible outcomes

- Has 9 possible genotypes
- Has 4 possible phenotypes

Independent Assortment: the principle states that genes for different traits can segregate independently during the formation of gametes

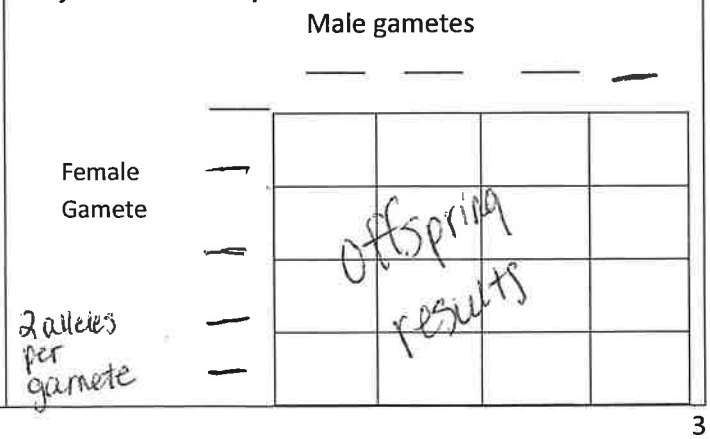
How to do a Punnett Square – we will do this together but here is the basic set-up
Refer to our Pea Chart Handout or Page 316 in your textbook

Monohybrid Punnett Square



* 1 allele per gamete

Dihybrid Punnett Square



Incomplete Dominance:

The heterozygote phenotype = mix

Situation where 1 allele is not completely dominant over another

- Both alleles are expressed in the heterozygous individual
- The heterozygous phenotype is a mix of the 2 alleles
- Example a Snapdragon Flower
  - C^RC^R = flower is red
  - C^WC^W = flower is white
  - C^RC^W = flower is pink
- Other examples include:

Codominance:

The heterozygote phenotype = Both shown

situation where the phenotype of the heterozygote shows both elements of same time

- The heterozygous phenotype shows Both alleles
- Example Roan coat of a horse/cattle
  - C^RC^R = horse is red
  - C^WC^W = horse is white
  - C^RC^W = horse is Roan
- Other examples include: Chickens, blood type

Multiple Alleles: A gene that has more than 2 alleles

- Example: Blood (A, B, O, AB); rabbit coat
- Each organism gets a total of 2 alleles (one from each parent) but there are more than 2 to choose from

Polygenic Trait: when traits are controlled by the interaction of 2 or more genes

- Examples: skin color, hair color, eye color

~Genes and the environments~

Environmental conditions can affect gene expression and influence genetically determined traits

- Examples: butterfly, Siamese cats, flowers

Chapter 14 Human Heredity Page 391

How do we "see" our chromosomes?"

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

- Scientists look at Chromosomes to study the genome
- Chromosomes are best seen in metaphase (mitosis) since they are in the middle of the cell
- The chromosomes are cut and placed into a picture called a Karyotype
- Karyotype: a diagram that shows the complete diploid set of chromosomes grouped together in pairs
  - It is arranged in order of decreasing size of chromosomes
  - Humans have 46 total chromosomes
  - Humans have 23 pairs of chromosomes
  - The Chromosomes are aligned up in pairs
  - The last set or the 23rd pair are called the sex chromosomes
    - These determine the sex of an organism
    - Males have XY sex chromosomes
    - Females have XX sex chromosomes
  - Set numbers 1-22 are called Autosomes
    - These determine the rest of an organisms' traits

Carrier: an individual who is heterozygous for a trait

- Has one dominant allele that covers/masks one recessive allele
- This individual does not show the recessive trait since the dominant has over powered it
- They are "carrying" it and can possible pass it on to the next generation/offspring

Sex-Linked Traits

- Traits controlled by genes located on the X chromosome
- Males are affected most often since they have only one X chromosome
- Females are less likely to express the trait since they have two X chromosomes
- Examples include: color-blindness, hemophilia

Chromosome Inactivation

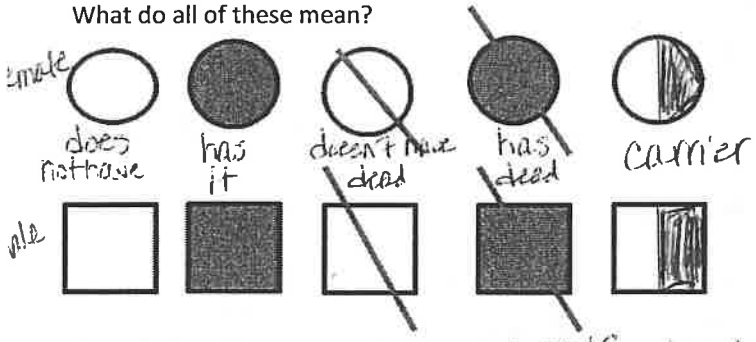
- Females have 2 dosages since have two X chromosomes
- The X chromosome is necessary for development of both males and females
- One of the X female chromosomes will stop functioning in all of the females cells
  - This creates a Barr Body (dense region in a female nucleus)
- The Y chromosome of males continues to work to determine male characteristics
- This can affect the coat color of some cats (Calico)
  - Black color means it has "Black" on the X sex chromosomes and can be either male/female
  - Orange color means it has Orange on the X sex chromosomes and can be either male/female
  - Calico means it has 1 Black & 1 Orange on X sex chromosomes and can be only female

Practice

How do you demonstrate how a particular trait is passed on in a family?

Pedigree: a chart that shows the pattern of inheritance for a particular trait within a family

What do all of these mean?



marriage

offspring

siblings

Roman Numerals= generation #

Arabic Numerals= individuals within a generation

Dominant pedigree demonstrates = male/female who have it in every generation

Recessive pedigree demonstrates= skips a generation

Autosomal Pedigree shows: trait on chromosome #1-22

Sex-Linked Pedigree shows: trait on X chromosome (usually males get it)

~Human Genetic Disorders~

How can a change in DNA affect a human's traits?

Changes in a gene's DNA sequence can change proteins by altering their sequences which directly affects their phenotype

Genetic Disorders

Disorder	Cause	Effect	Population	Dominant/ Recessive	Autosomal/ Sex-Linked
Cystic Fibrosis	defective membrane protein	White ↔ build-up of thick mucus in lungs	European	recessive	autosomal
Sickle Cell Disease	defective RBC (hemaglobin)	sickle shaped RBC	African American	recessive	Autosomal
Huntington's Disease	extra protein in brain	nervous system deteriorates	middle age	dominant	Autosomal
Tay-Sachs Disease	absence of enzymes to break fatty substance in brain	fat deposits build-up on brain	Jewish	recessive	Autosomal
Color Blindness	defect in cones of eyes	cannot see color	male	recessive	sex-linked
Hemophilia	gene defect in blood clotting	cannot stop bleeding	male	recessive	sex-linked
Achondroplasia	a gene affecting bone growth	dwarfism	All	dominant	autosomal

Genetic Advantages

Malaria Heterozygote

- NN - normal
- NS - normal - resistant to malaria
- SS - sickle cell

Cystic fibrosis heterozygote

- CC - normal
- Cc - normal — resistant to Typhoid fever
- cc - cystic fibrosis

Chromosomal Disorders

- This means there is an error in meiosis
- Each human gamete should have 23 chromosomes
- But what happens if the homologous chromosomes do NOT separate correctly
  - This is called NON-DISJUNCTION
    - A gamete with an abnormal number of chromosomes leading to a disorder of chromosome number
    - The gamete could end of having only 1 copy of an allele
      - Example: Turner's Syndrome
        - Females who have only 1 X chromosome
    - The gamete could end of having 3 copies of an allele
      - This is called Trisomy
        - Example: Down Syndrome
          - They have 3 copies of chromosome number 21
        - Example: Klinefelter's Syndrome
          - A male who has XXY sex chromosomes

Studying the Human Genome

- Human Genome Project
  - A 13 year international effort to sequence all of the 3 billion base pairs of human DNA which would identify all human genes
  - Complete in 2003
  - Helps us to locate disorders and diseases
  - Someone can know their exact genetic make-up and what they can pass on to their offspring