Chapter 1 ~ Mendel's First Law & Meiosis

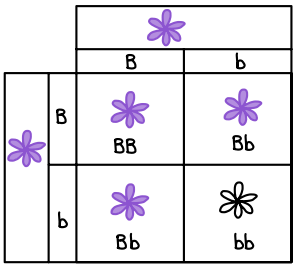
Introduction:

Gregor Mendel
- Augustinian Monk
- Plant Breeder
  - ↳ Quantitative study of heredity

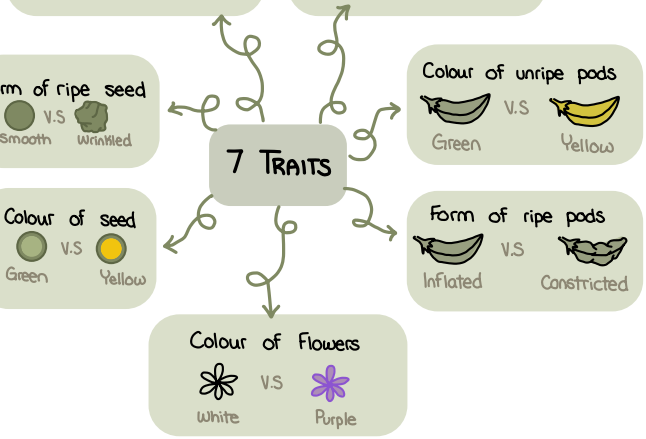
Experiments...
- Garden Pea Plants

Why Peas?
1. True Breeding
2. Display variety of contrasting traits
3. Reproduce via self-pollination
4. Grow quickly & require minimal space

Alleles
- Single trait can exist in different versions even within an individual plant / animal

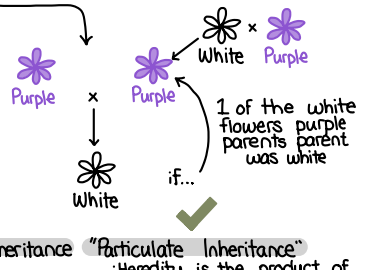


7 Traits



8yrs of experiments later...

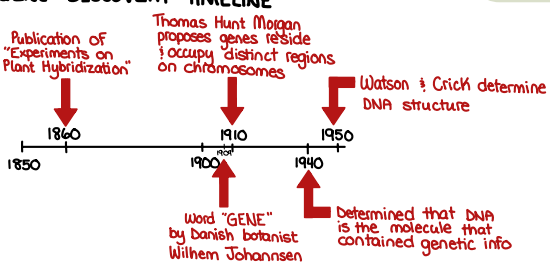
Foundational Principles of Inheritance
1. The Law of Equal Segregation
2. The Law of Dominance
3. The Law of Independent Assortment



✖ Blending Inheritance

✔ "Particulate Inheritance"
- Heredity is the product of discrete factor that controls independent traits

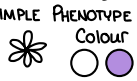
Gene Discovery Timeline



Law of Equal Segregation
- During gamete formation two alleles at a gene locus segregate from each other & each gamete has an equal probability of containing either allele

Dominant & Recessive Alleles

Heterozygous Phenotype
- Dominant - Recessive character is a relationship between two alleles
  - ↳ MUST be determined by observation of the heterozygous phenotype
- Simple Phenotype Example:
  - Colour



Determine via Observations...

Homozygous bb or BB vs **Heterozygous** Bb

This allows us to determine that... *Alleles are DIFFERENT versions of a gene

- ○ allele = Recessive
  - It's phenotype is masked by purple allele in a heterozygote
- ● allele = Dominant
  - It's phenotype is ALWAYS displayed over white allele so in a heterozygote a white coloured flower is produced

Written Form
- b = Recessive allele (lowercase)
- B = Dominant allele (Capital)
- Alleles of the SAME gene write with SAME letter
- Genes & alleles are written in *italics*
- Proteins are written in ALL CAPITALS!
  - ↳ ex: white gene (w) = WHITE

Different Types of Allele Relationships
- Complete Dominance
- Incomplete Dominance
- Co-Dominance

Note: Mendel studied trait with his peas were all COMPLETELY dominant

In Diploid Organism...
- For a dominance allele only ONE copy of that allele is needed to express the dominant phenotype
- For a recessive allele the gene NEEDS to have TWO copies (or be homozygous) to express the recessive phenotype

Examples of Symboles used to Represent Genes & Alleles

Alleles	Meanings
A & a	Uppercase letters represent dominant alleles & lowercase letters indicate recessive alleles. Mendel invented this system but it is NOT commonly used because not all alleles show complete dominance & many genes have more than 2 alleles.
a^+ & a_1	Superscript or subscripts are used to indicate alleles. For wild type alleles the symbole is a superscripte "+".
AA or A/A	Sometimes a forward slash is used to indicate that the two symboles are alleles of the same gene, but on homologous chromosomes

MEIOSIS

STAGES OF MEIOSIS

- Eukaryotic ← Reproduce SEXUALLY

Single celled eukaryotes that undergo meiosis are called MEIOCYTES (2n)

1 cell + 1 cell = OFFSPRING

FOR SUCCESS... cells that fuse MUST contain HALF the number of chromosomes as in the ADULT organism

if not... Number of chromosomes would double w/ each generation, which would be UNSUSTAINABLE

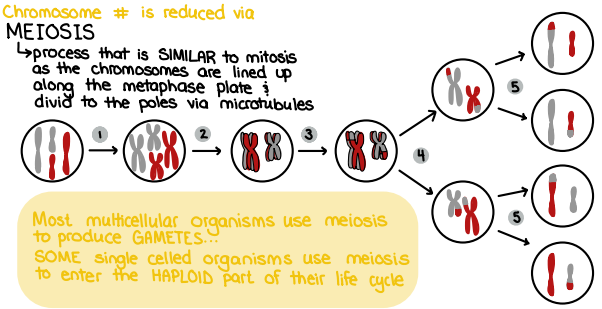
MEIOSIS
- I: Homologous Chromosomes SEPERATE
- II: Homologous Chromosomes SEGREGATE

Reductional division
- Because # of chromosomes inherited in each of the daughter cells
- Parent cell = 2n
- 2 daughter cells = 1n

Further divided into
- Prophase I
- Metaphase I
- Anaphase I
- Telophase I

When DNA repair enzymes break the DNA of two non-sister chromatids together to create a cross over b/w 2 non-sister chromatids

Chromosome # is reduced via MEIOSIS
↳process that is SIMILAR to mitosis as the chromosomes are lined up along the metaphase plate & divid to the poles via microtubules



Most multicellular organisms use meiosis to produce GAMETES...
SOME single celled organisms use meiosis to enter the HAPLOID part of their life cycle

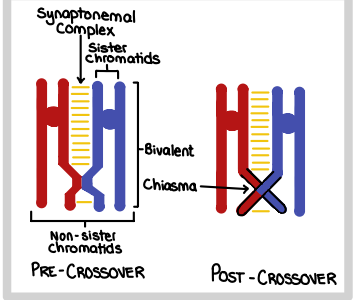
MEIOSIS

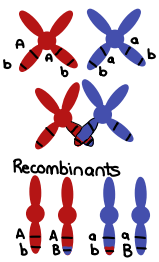
- Begins similairly to mitosis in that a cell has grown large enough to divide & has replicated it's chromosome
- Differs from mitosis because it requires two rounds of division
  - Sister chromatids segregate in meiosis II
- In meiosis I prophase & metaphase I homologous chromosomes pair up. "Synapse" and are called BIVALENTS
  - ↳In mitosis homologous chromosomes LINE up individually during metaphase

KEY DIFFERENCE

IMPORTANT:
- This difference affects the segregation of alleles, and allows for recombination to occur through cross-over

CROSS-OVERS...

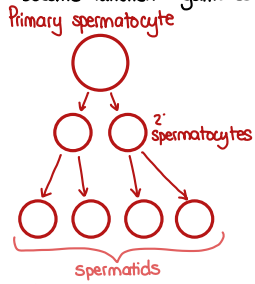




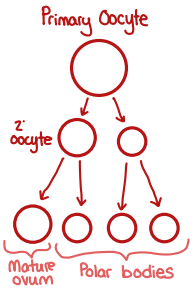
*Cross overs function to HOLD homologous chromosomes together during meiosis I so they can orient correctly & segregate successfully
↳it also reshuffles allele combinations along chromosomes = GENETIC DIVERSITY

GENE MATURATION

In plants & animals...
- Cell by meiosis need to MATURE before they become functional gametes

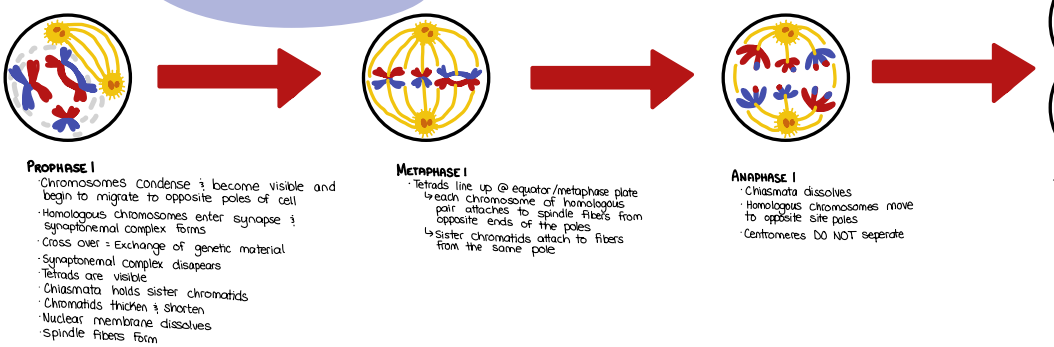


♂ Animals
- 4 spermatids w/ tails (sperm)



♀ Animals
- Gamete = eggs
- For max nutrients only one of four meiosis products become an egg
- Other 3 products end up as disposable cells called polar bodies

products of meiosis reproduce a few times via mitosis as they develope into functional ♀/♂ gametes

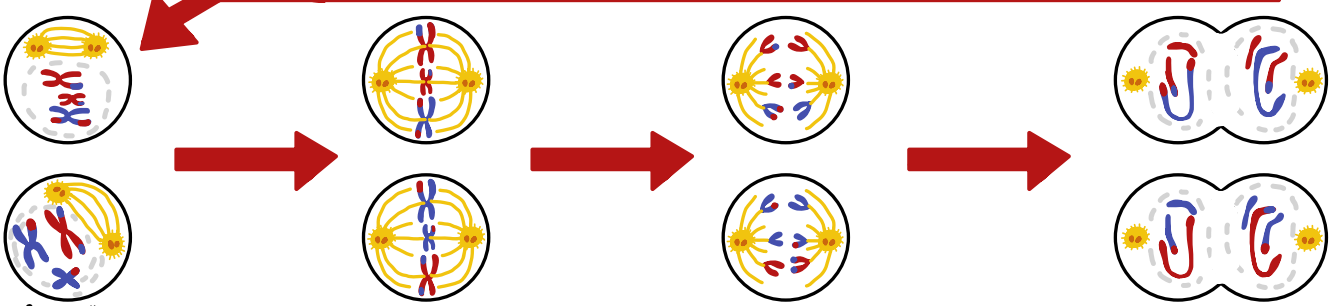


PROPHASE I
- Chromosomes condense & become visible and begin to migrate to opposite poles of cell
- Homologous chromosomes enter synapse & synaptonemal complex forms
- Cross over = Exchange of genetic material
- Synaptonemal complex disapears
- Tetrads are visible
- Chiasmata holds sister chromatids
- Chromatids thicken & shorten
- Nuclear membrane dissolves
- Spindle fibers form

METAPHASE I
- Tetrads line up @ equator/metaphase plate
  - ↳each chromosome of homologous pair attaches to spindle fibers from opposite ends of the poles
  - ↳Sister chromatids attach to fibers from the same pole

ANAPHASE I
- Chiasmata dissolves
- Homologous chromosomes move to opposite site poles
- Centromeres DO NOT seperate

TELOPHASE I & CYTOKINESIS
- Nuclear envelope reforms
  - ↳Resulting cells have ½ # of chromosomes each w/ 2 sister chromatids
- Similar to interphase but NO chromosome duplication occurs
- Daughter nuclei become enclosed into seperate daughter cells



PROPHASE II
- Chromosome condense
- Centrioles move towards the poles
- Nuclear envelope disintegrates

METAPHASE II
- Chromosomes align @ equator/metaphase plate
- Sister chromatids attach to spindle fibres from opposite poles

ANAPHASE II
- Centromeres divid
- Sister chromatids move to opposite poles

TELOPHASE II & CYTOKINESIS
- Chromosomes begin to uncoil
- Nuclear envelope & nucleoli begin to reform
- Division of the cytoplasm occurs
  - ↳Forms four new daughter cells each containing haploid number of chromosomes

PUNNET SQUARES

Named after R.C. Punnett

GREAT TOOL FOR CALCULATING EXPECTED GENOTYPIC & PHENOTYPIC RATIOS

- The specific ratios seen in monohybrid crosses can be described using a punnett square
- Given the GENOTYPE of any two parents we can predict ALL possible genotypes of the offspring

NOTE: You can also determine the dominance relationship of all alleles
↳Allows us to predict offspring PHENOTYPE

Ex: Punnett Square of F_1

♂ \ ♀	A	a
A	AA	Aa
a	Aa	aa

3 Different Genotypes
- AA
- Aa
- aa

2 Different Phenotypes
- Dominant ← AA or Aa
- Recessive ← aa

RATIOS...
Genotypic → 1 : 2 : 1

Phenotypic → 3 : 1
- Dominant
- Recessive

GEN 1 CROSS: AA × aa

	a	a
A	Aa	Aa
A	Aa	Aa

100% dominant

GEN 2 CROSS: Aa × Aa

	A	a
A	AA	Aa
a	Aa	aa

75% AA or Aa (dominant)
5% aa (recessive)

1 Locus on a Chromosome - Segregation - Monohybride

Solved the mystery of inheritence as units (genes)

Also invented several testing and analysis techniques still used today

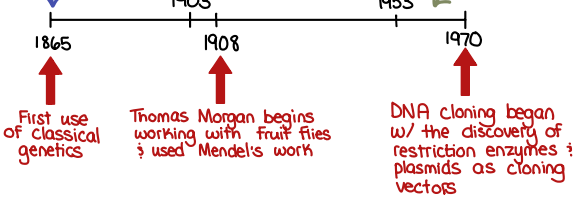
Classical Genetics

- Science of examining biological questions using CONTROLLED matings of model organisms

Widespread use after rediscovery by:
- E. von Tschermak
- H. de Vries
- C. Correns
- W.J Spillman

Molecular Genetics

- Science of solving biological problems using DNA & RNA and proteins



Terminology

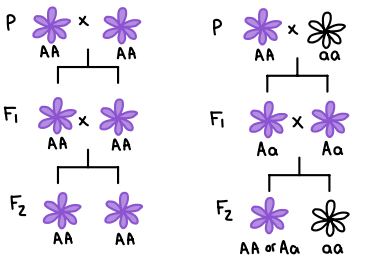
- **Locus** - A specific position, region, or segment along a chromosome.
  - ↳Each gene occupies a specific locus
  - ↳Each locus will have an allelic form (allele)
- **Genotype** - The complete set of alleles (at all loci of interest) in an individual
  - ↳When writting out a genotype, only alleles @ loci of interest are considered, the rest are considered wild type & not written
- **Phenotype** - The observable/detectable effect of alleles on the structure/function of that individual
  - ↳May range from simple, visible traits (Hair colour) to complex ones (disease susceptibility / behaviour)

True-Breeding Lines

- In-bred populations in which all parents & their offspring (over MANY generations) have the SAME phenotype w/ respect to a particular trait

*Useful because they are typically assumed to be homozygous for the alleles that affect the trait of interest

Two individuals (homozygous) are crossed all their offspring will ALSO be homozygous
- ↳Continuation of such crosses constitutes a TRUE breeding line or strain



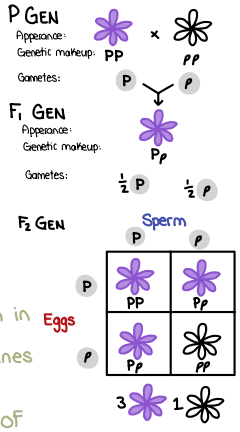
Monohybrid Cross

- One in which BOTH parents are HETEROZYGOUS (hybrid) for a single (mono) trait
  - ↳ex: petal colour in pea plants

Using monohybrid crosses Mendel discovered that genes were DISCRETE units that seperated in the creating of offspring

Previous = Blending Inheritance
- Would have meant that a cross b/w a white flower & a purple flower would create a blended phenotype

Instead Mendel saw...
- Distinct parental colours in the hybrids which creates specific ratios the purple & white seen in the parents
  - ↳Traits were NOT blended in true-breeding lines but instead parent alleles were carried on through offspring
- He discerned DOMINANT & RECESSIVE alleles of each trait in the studied pea plants



Recall: Law of Segregation
- States that every individual contains a pair of alleles for each gene, which segregate during the formation of gametes and so for every gene pair each parent passes on a random allele to it's offspring

Single Locus Test Cross

- Genotypes cannot be observed directly MUST be inferred based on phenotypes

Because of DOMINANCE it is hard to tell apart

Homozygous: AA & aa

Heterozygous: Aa

Results depend on genotype of Purple flower pea plant

To determine genotype of a given individual we perform a TEST CROSS!

Unknown genotype x aa (Homozygous recessive)

ex: w/ A_ x w/ (aa)

Purple flower means the genotype MUST contain at least 1 "A"

	A	A
a	Aa	Aa
a	Aa	Aa

100% Aa offspring if unknown is homo

	A	a
a	Aa	aa
a	Aa	aa

50% Aa & aa if unknown part is hetero