FRST302: Forest Genetics

Lecture 1.3: DNA Structure

Lecture 1 - Recap

- Mendel's laws and Classical Genetics
- Mechanisms of Mendel's laws
- From discrete particles to continuous variation
- Chromosomes

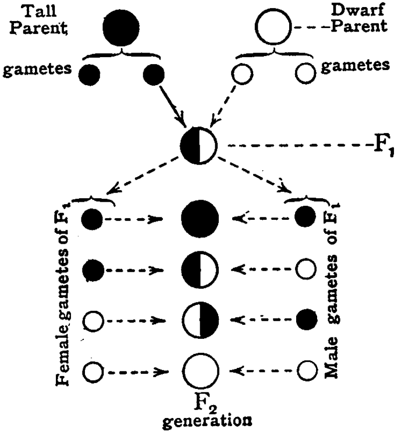


Figure from Punnett 1911

Outline for Today

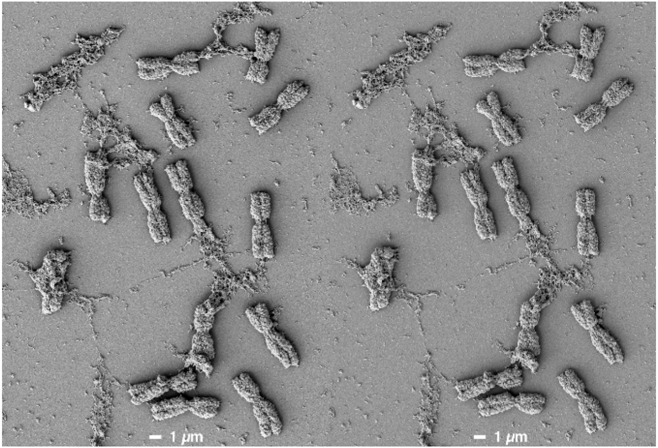
Chromosomes and Their Structure

Linkage & Genetic Mapping

DNA

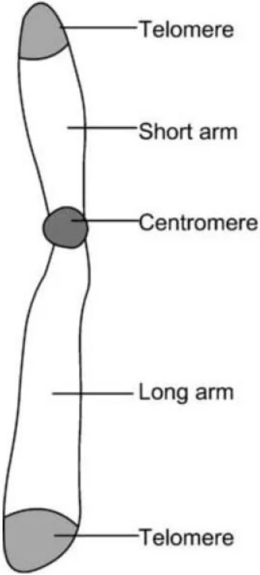
Chromosomes

Chromsosomes are the “particles of inheritence”,
but what are they?

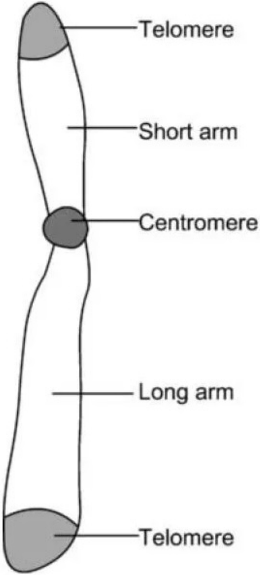


SEM of barley chromosomes in metaphase: Schroeder-Reiter and Wanne 2013 *SEM for the Life Sciences*

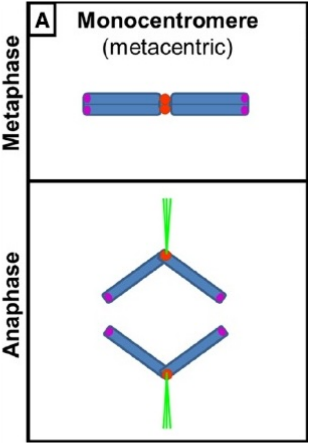
Chromosome Structure



Chromosome Structure



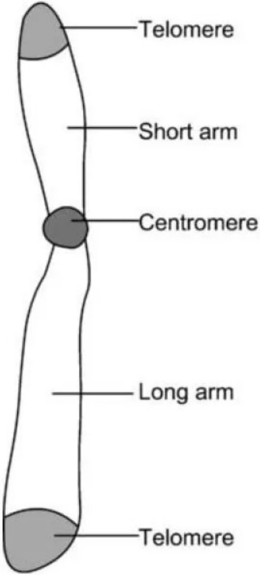
Centromeres play a structural in meiosis

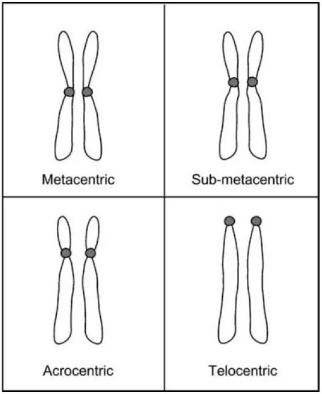


[a]

[a] Modified from *Cuacos et al 2015 Front. Plant Sci.*

Chromosome Types





Chromosome Numbers

The number of sets of homologous chromosome organisms possess varies widely!

Humans	23
Maize	10
Banana	11
Loblolly pine	12

There is no known correlation between organisms complexity and chromosome count

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Douglas-fir has 13 chromosomes, but recently underwent a chromsome fission

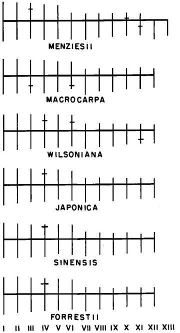
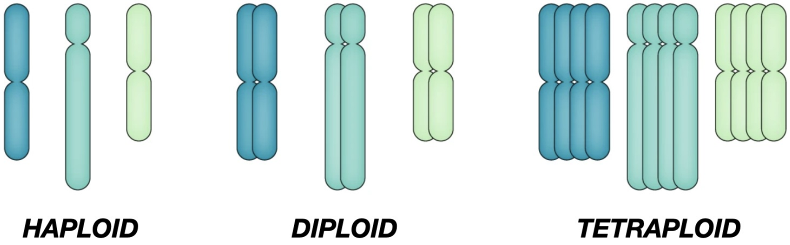


FIGURE 1. *Idiograms of six species of the genus Pseudotsuga. Cross marks are secondary constrictions. Idiograms of P. menziesii and P. wilsoniana from Thomas and Ching (1968). Idiogram of P. macrocarpa from Christiansen (1963).* [a]

[a] Idiogram from Doerksen and Ching 1972 *Forest Science*

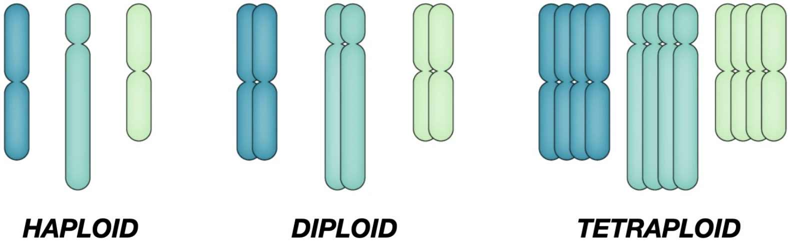
Chromosomes - Ploidy



Ploidy refers to thenumber of homologous chromosome copies an organism possesses

Differences in ploidy are extremely common in plants.

Chromosomes - Ploidy



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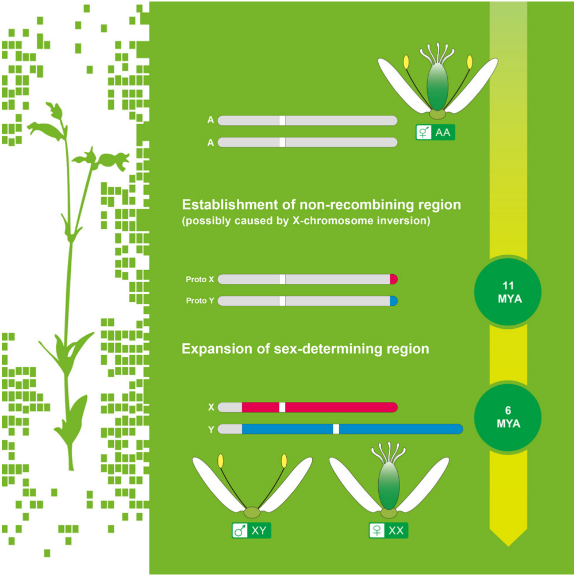
Differences in ploidy are extremely common in plants.

Pasta wheat is tetraploid (4n) and bread wheat is hexapoid (6n)!

Sex Chromosomes



Yue et al 2023 -*Current Biology*

In many organisms, specific chromosomes or chromosomal regions are linked to the expression of sex-specific traits.

Silene latifolia, for example, has an XY sex-chromosome system

Refresher...

The law of segregation: each individual possesses a pair of particles for any particular trait and each parent passes one of these randomly to its offspring

Mendel's Laws

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The law of dominance: for some traits, the presence of one kind of particle masks the presence of another. Mendel referred to the **dominant** particle as masking the effects of the **recessive** particle

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The law of independant assortment: when two individuals differ in more than two pairs of traits (e.g. smooth v. wrinkly and green v. yellow), the inheritance of one pair of traits is independent of another

Dihybrid Cross

In the last lecture, we restricted ourselves to looking at the expected ratios of genotypes for a single trait in a given cross, but there's no reason we need to do that

Let's now follow the inheritence of two traits instead and stick with smooth v. wrinkly and yellow v. green

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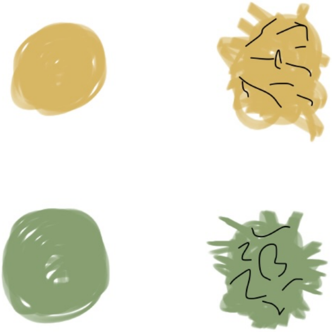


Dihybrid cross

Let's cross "true breeding" peas that were smooth and yellow with peas that are wrinkly and green

Parental Generation:  *x* 

Genotypes: YYWW *x* yyww

Let's cross "true breeding" peas that were smooth and yellow with peas that are wrinkly and green

Parental Generation:  x 

Genotypes: YYWW x yyww

↓

F1 Generation:  x 

Genotypes: **YyWw** x **YyWw**

Dihybrid Cross

If we filled out the Punnett square for this cross, what would the expected ratio of phenotypic combinations be?

YyWw

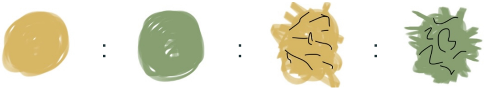
YyWw

	YW	Yw	yW	yw
YW				
Yw				
yW				
yw				

Dihybrid Cross

		YyWw			
		YW	Yw	yW	yw
YyWw	YW	YYWW	YYwW	yYWW	yYwW
	Yw	YYWw	YYww	yYWw	yYww
	yW	YyWW	YywW	yyWW	yywW
	yw	YyWw	Yyww	yyWw	yyww

In what proportions would we expect to see the different phenotypic combinations?



Dihybrid Cross

According to the **Law of Independent Assortment**, we would expect:

$9x$  : $3x$  : $3x$  : $1x$ 

Results of a Dihybrid Cross in Sweetpeas

In the early 1900s, Bateson and Saunders conducted a series of experiments using sweet peas (*Lathyrus odoratus* - not garden peas like Mendel)

Rather than seed colour and texture, they were examining flower colour and the shape of pollen grains

They conducted a dihybrid cross and got the following results:

Phenotype	**Observed**
Purple, long	1528
Purple, round	106
Red, long	117
Red, round	381
Total	2132

Results of a Diybrid Cross in Sweetpeas

With 2132 plants and an expected phenotypic proportions of 9 : 3 : 3 : 1, we can quantify how strange the deviations from the expectations are

Phenotype	**Expected**	**Observed**	
Purple, long	1199	1528	
Purple, round	400	106	
Red, long	400	117	
Red, round	133	381	
Total	2132	2132	

Results from: *Bateson, W., Saunders et al. Experimental studies in the physiology of heredity. Reports to the Evolution Committee of the Royal Society 2, 1–55, 80–99 (1905)*

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Phenotype	**Expected**	**Observed**	**(Obs.-Exp.)^2/Exp.**
Purple, long	1199	1528	90.3
Purple, round	400	106	216.1
Red, long	400	117	202.2
Red, round	133	381	462.4
Total	2132	2132	$\chi^2 = 969.0$

This χ^2 test gives a $p-$value < 0.0001

Results from: *Bateson, W., Saunders et al. Experimental studies in the physiology of heredity. Reports to the Evolution Committee of the Royal Society 2, 1–55, 80–99 (1905)*

Q: Why would we see deviations from the Law of Independant Assortment?

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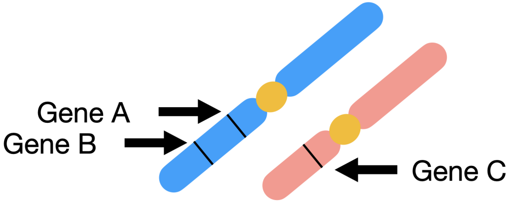
A: Linkage!

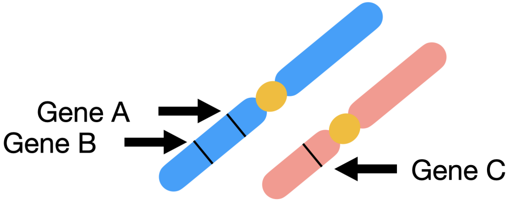
Genetic Linkage



– The law of the independent assortment was derived based on genes located **on different chromosomes**. Alleles of these genes **segregate independently** during meiosis

Figure from: *Brown 2002*

Genetic Linkage



- The law of the independent assortment was derived based on genes located **on different chromosomes**. Alleles of these genes **segregate independently** during meiosis
- Genes located on the same chromosome may be inherited together during meiosis

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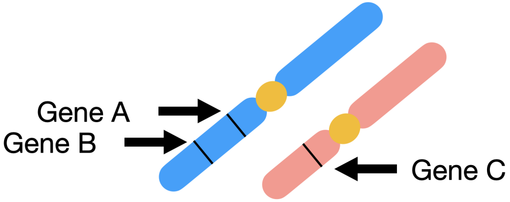
Genetic Linkage



- The law of the independent assortment was derived based on genes located **on different chromosomes**. Alleles of these genes **segregate independently** during meiosis
- Genes located on the same chromosome may be inherited together during meiosis
- But this is inadequate to explain the emergence of new trait combinations in the sweet peas

Figure from: *Brown 2002*

Crossing Over

- In the 1910s, Thomas Hunt Morgan developed genetic experiments with the fruit fly *Drosophila melanogaster*
- Morgan and colleagues observed cases where expected ratios for linked factors broke down (just like with the sweet peas)
- They proposed a process of "*crossing-over*" where alleles may swap onto alternate chromosome pairs

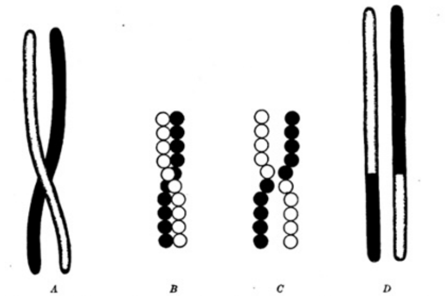


FIG. 24.—Diagram to represent crossing over. At the level where the black and the white rod cross in *A*, they fuse and unite as shown in *D*. The details of the crossing over are shown in *B* and *C*.

Image from *Morgan, T.H., Sturtevant, A.H., and Bridges, C.B. (1915). The Mechanism of Mendelian heredity.*

Linkage in Sweet Peas

Phenotype	Observed
Purple, long	1528
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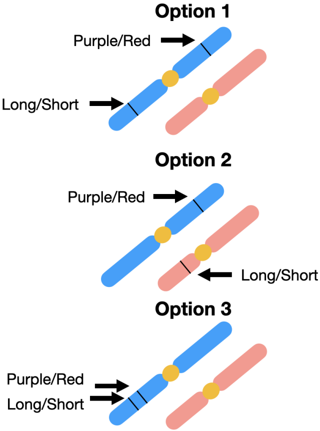
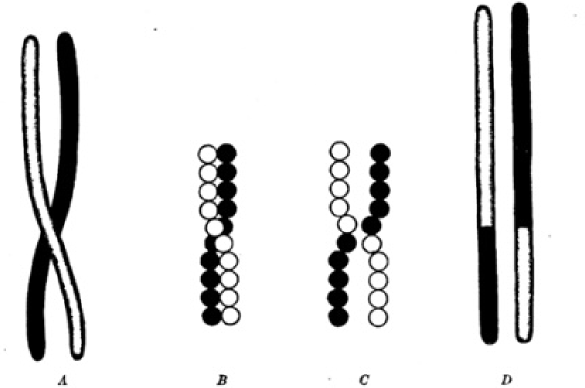
Which of the following do you think is the closest approximation of the arrangement of genes in the sweetpea?



Crossing Over



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Measuring Genetic Distance

If we assume that the purple/red and long/round genes are on the same chromosome, we can calculate the genetic distance between them

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Number of recombinant genotypes $= 106 + 117$
Recombination fraction $= 0.105$

We usually express these recombination units as:
$100 \times RecombinationFraction = 10.5cM$

Where cM stands for centimorgans: the expected number of recombination events observed in 100 meioses

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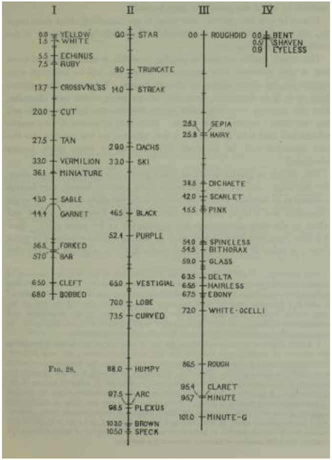
What is the maximum genetic distance possible between two markers?

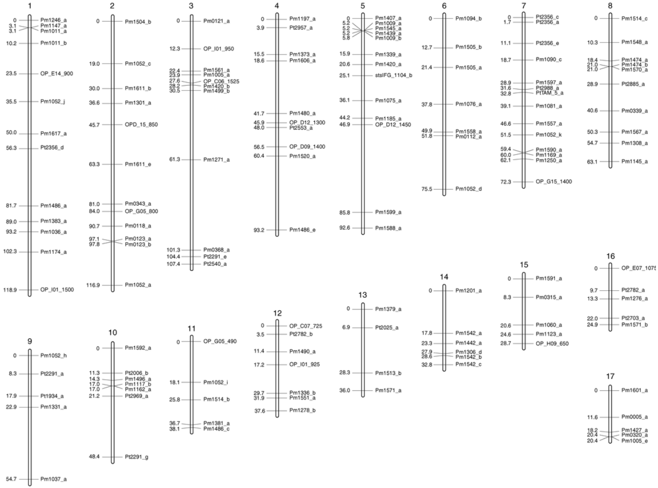


- The frequency of “crossed-over” trait combinations gave Morgan and colleagues the ability to identify the order of genes on the *D. melanogaster* chromosomes
- More amazingly, they had the insight that the relative frequency of cross-overs could be used to quantify the distance between genes along the chromosomes

Morgan 1922

Douglas-fir Genetic Map

These principles are the basis of **genetic mapping**, a technique that is still widely used today

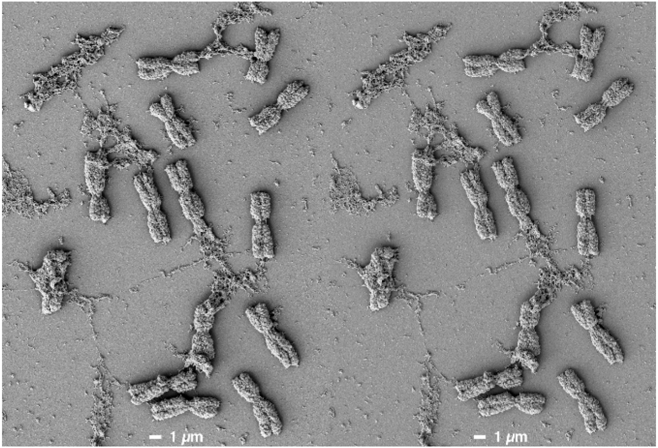


Though we now use molecular markers rather than traits

Questions?

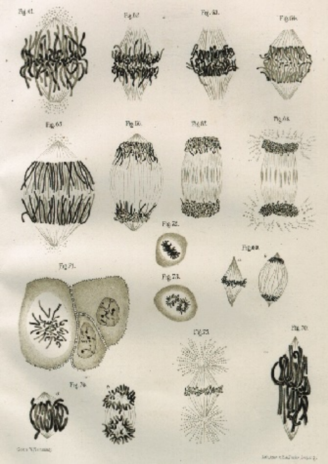
Questions?
Let's take a short break

Chromsosomes are the "particles of inheritence", but what are they made of?



SEM of barley chromosomes in metaphase: Schroeder-Reiter and Wanne 2013 *SEM for the Life Sciences*

A Timeline of Some Discoveries



1865 Mendel postulates laws of inheritance

1869 DNA Isolated - though it was unclear what its relevence was unclear

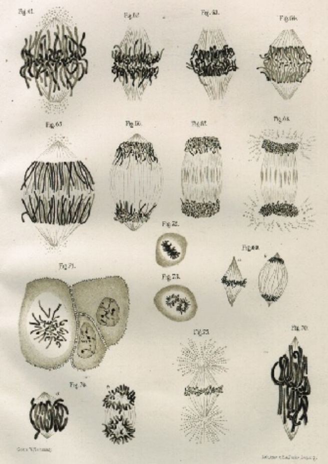
1882 Discovery of the fibrous network of "chromatin" (*stainable material*) and chromosomes within nuclei

1902-6 Sutton-Boveri chromosome theory - the segregation of chromosomes during meiosis matches the segregation pattern of Mendel's laws

1915 Morgan demonstrated that chromosomes carry genes, and also discovered genetic linkage *won Nobel Prize in 1933*

Drawing of mitosis by Walther Flemming 1882

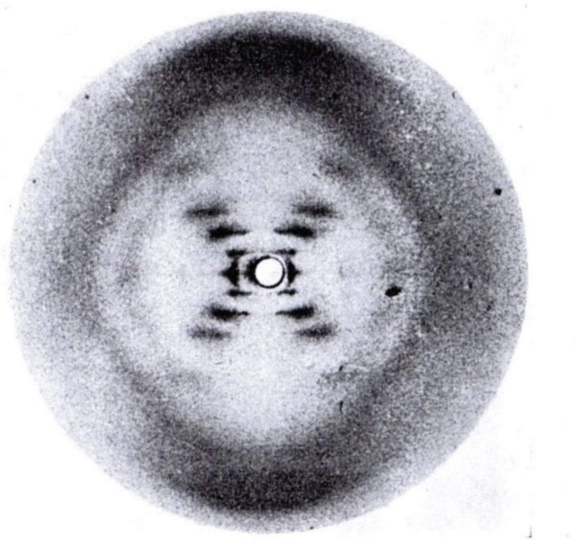
A Timeline of Some Discoveries



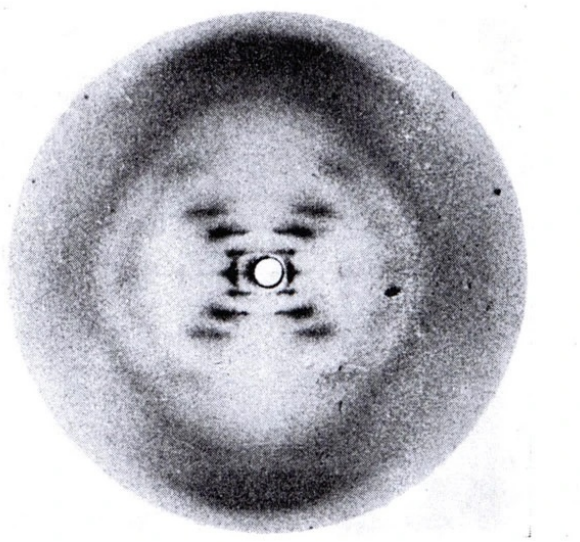
- 1915 Morgan and Sturtevant constructed their genetic map for *D. melanogaster*
- 1932 Barbara McClintock confirms that genes are exchanged during crossing-over
- 1940s DNA is determined to be the material within chromosomes that carry heritable information
- 1950 The composition of DNA is determined - including Chargaff's rules

Drawing of mitosis by Walther Flemming 1882

Photo 51



1952-1953 - The Structure of DNA is Determined



This photograph, captured by Rosalind Franklin, led directly to the discovery of the structure of DNA

The Structure of DNA

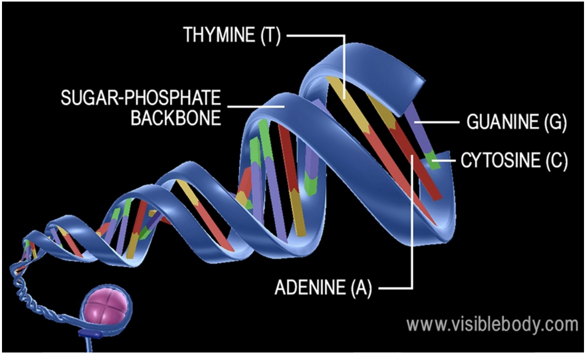
- A molecule of DNA has two strands that form a double helix shape structure, like a twisted ladder
- Each step on the "ladder" is made up of a pair of nitrogenous bases, that are designated with the letters:

  A – adenine

  C – cytosine

  G – guanine

  T - thymine



They are not really coloured!

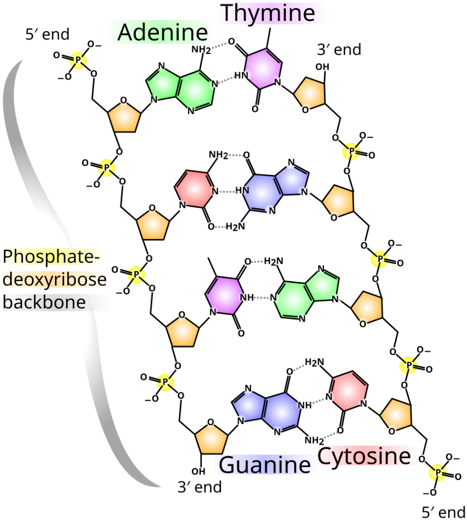
The Structure of DNA

DNA is a polymer

- Polynucleotide – repeating units

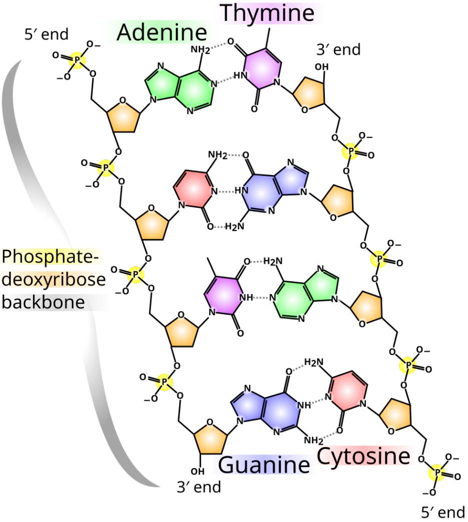
A nucleotide has 3 components:

- 5-carbon sugar (deoxyribose)
- Phosphate
- Nitrogenous base



The Structure of DNA

- DNA is a polymer
  - Polynucleotide – repeating units
- DNA is chiral
  - There is a differences between the 5' to 3' strands compared to the 3' to 5' strands



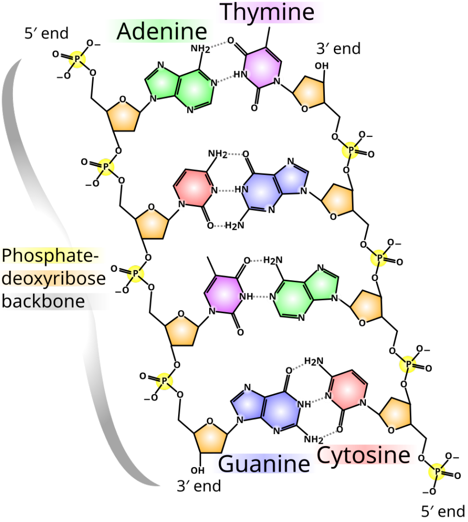
The Structure of DNA - Pairing Rules

A always pairs with **T**

C always pairs with **G**

Each rung on the "ladder" is referred to as a basepair

The DNA in the diagram is, for example, 4bp long



These pairing rules were first described by Chargaff - e.g. the number of Cytosine in a chromosome is equal to the number of Guanines

Genome

- The term "**Genome**" refers to the collection of all genetic material possessed by an organism
- It consists of DNA (and RNA[1]).
- The genome includes chromosomal DNA and other DNA held within cell
- For most organisms, a copy of the genome is held within each cell[2]

[1] see next lecture
[2] exceptions abound

Genome

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Any guesses for the total mass of DNA (in kg) in the human average human body?

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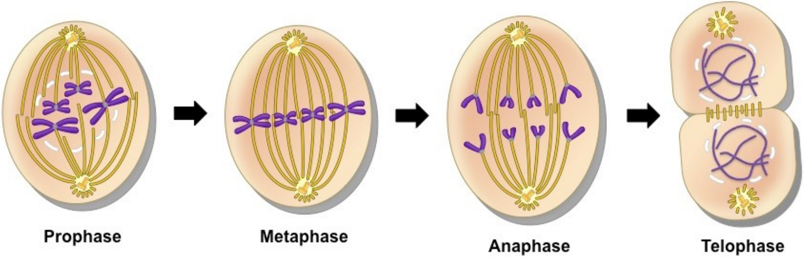
[1] see next lecture

[2] exceptions abound

Mitosis

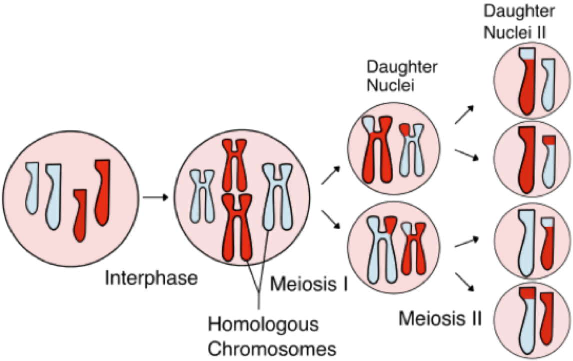


Mitosis is the division of a mother cell into two genetically identical copies

It is to increase the number of cells for growth in size

Image from: *Bioninjas*

Meiosis



Meiosis is the process by which gametes are formed

In a diploid organisms gametes are haploid cells[1]

Segregation, independent assortment and crossover have important implications for all aspects of life

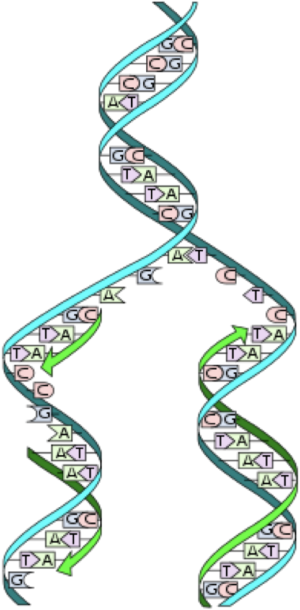
[1] In a tetraploid gametes are diploid
Image from: *Wikipedia*

Meiosis is Required for Sexual Reproduction

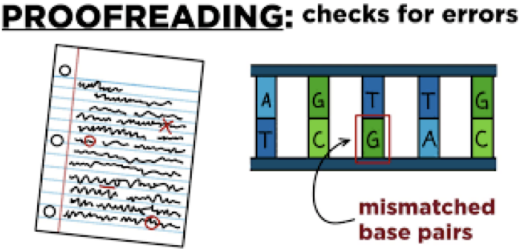
DNA Replication

- Both mitosis and meiosis involve DNA replication
- DNA replication produces two identical replicas of DNA from one original DNA molecule.
- During replication, the double-stranded DNAs are separated. Each strand of the original DNA molecule serves as a template for the production of its counterpart.
- This process occurs in all living organisms and is the basis for growth and inheritance. [a]

[a] Image from: *Wikipedia*



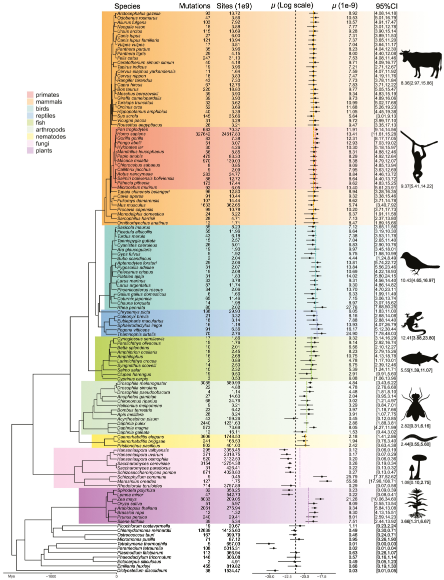
DNA Replication Errors



Errors in the DNA replication process are rare (about 1 in 100,000 basepairs) - mismatch repair catches them

When errors are missed, they become mutations

Mutation Rates



[a]

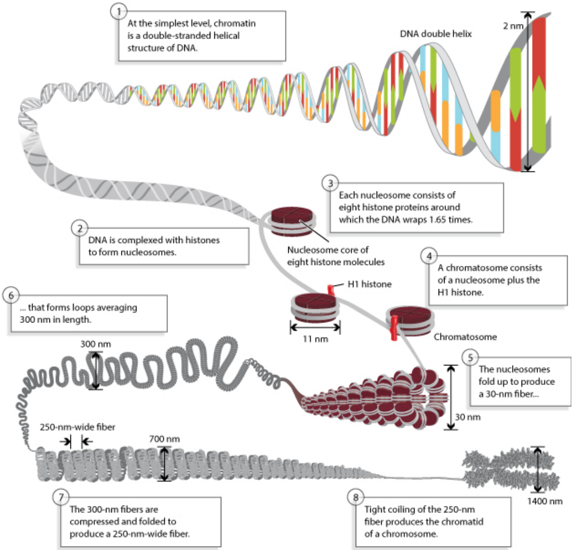
Mutations are the ultimate source of all genetic variation

The mutation rate in eukaryotes hovers around $1 \times 10^{-9}/bp$

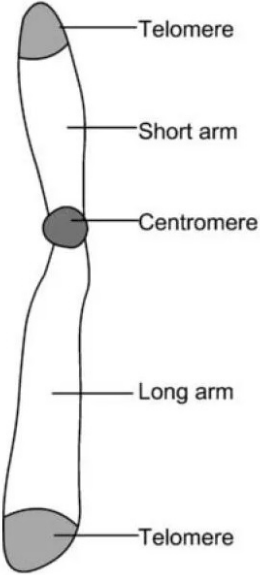
This is a small number, but genomes are big (we'll discuss this again in a subsequent lecture)

[a] Wang and Obbard 2023 *Evol. Letters*

How is DNA stored?



Chromosome Structure - Telomeres

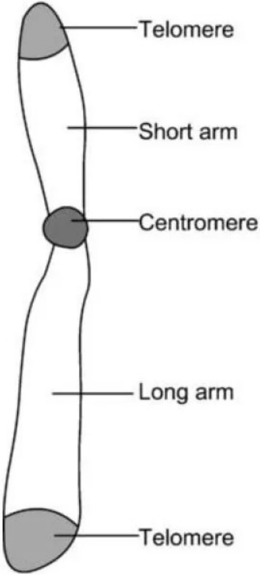


Telomeres

The "shoelaces" of the chromosome, telomores protects the ends of chromosomes from damage and from fusing with others

In plants, the telomere consists of a sequence TTAGGG repeated many thousands of times

Chromosome Structure - Centromeres



Centromeres

The “belt” of the chromosome, centromeres are compsed of specifc DNA repeats

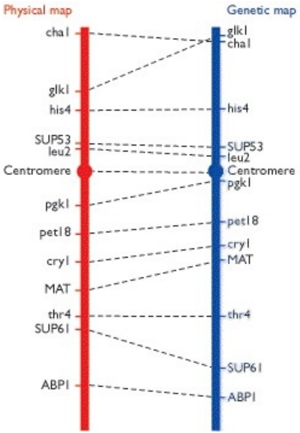
Centromeres recruit enzymes necessary for cell division protects the ends of chromosomes from damage and from fusing with others

Learning Outcomes

- Chromosome structure
- Genetic linkage
- Genetic mapping
- DNA structure
- Mutation

A Question to Think About...

Here's a comparison of a genetic map in yeast *Saccharomyces cerevisiae* with an estimate of the physical map (where the genes sit on the DNA itself)



Why would the distances on the genetic map and the physical map differ?

Recommended Reading/Viewing

All recommended reading is openly available and accessible online

Reading

Chapter 5 of *Genomes* by T. Brown.
https://www.ncbi.nlm.nih.gov/books/NBK21116/

Viewing - both are fairly short

DNA Structure: https://youtu.be/C1CRrtkWwu0
DNA Replication: https://youtu.be/TNKWgcFPHqw