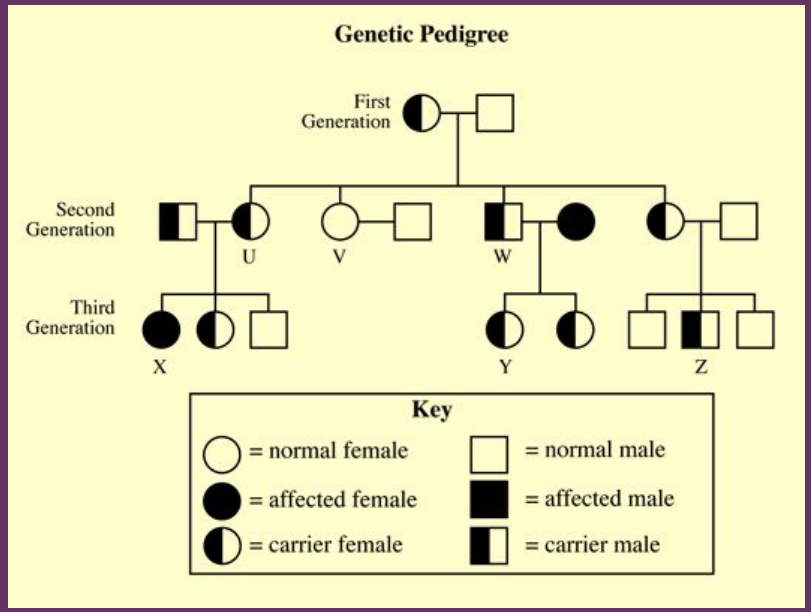
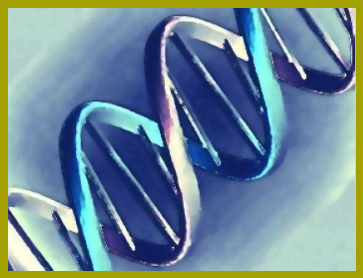
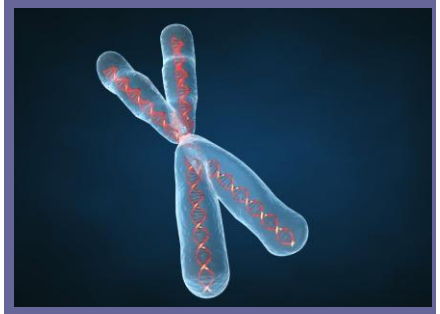
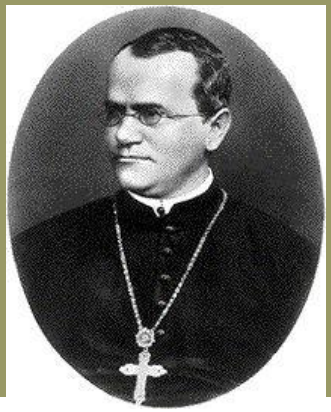

Genetic Pedigree
First Generation
Second Generation
Third Generation
U
V
W
X
Y
Z
Key
= normal female
= affected female
= carrier female
= normal male
= affected male
= carrier male





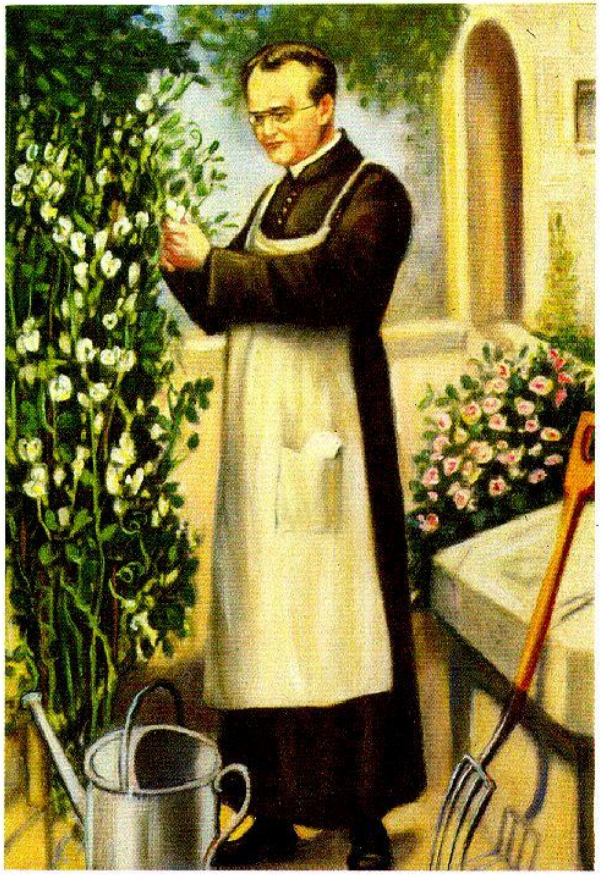


Genetics

What is Genetics?

- Genetics is the scientific study of **heredity**, which refers to the passing of characteristics from parents to offspring.

Gregor Mendel

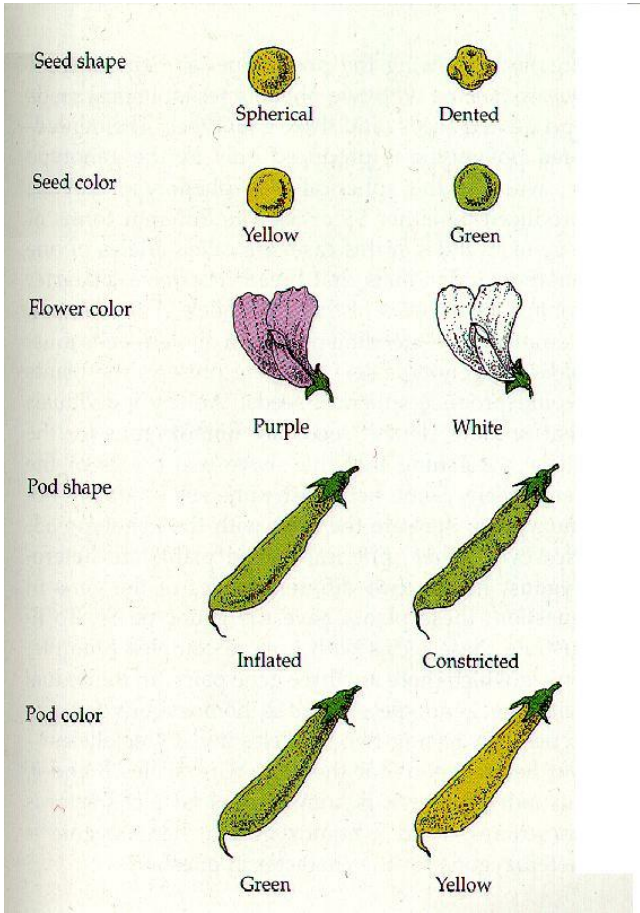


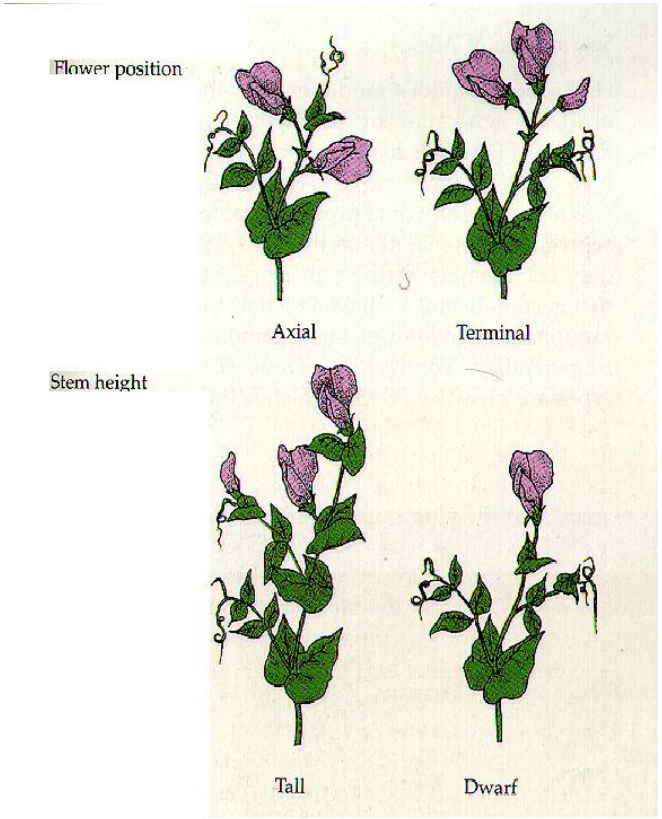
Gregor Mendel

- The "Father of Modern Genetics"
- An Austrian monk who worked in a monastery garden in the mid 19th century.
- Grew and studied pea plants—wondered why different pea plants had different characteristics (tall vs. short, green vs. yellow seeds, white vs. purple flowers, etc).
- Experimented with thousands of pea plants to better understand heredity; this work forms the basis of modern genetics.

Mendel's Experiments with Pea Plants

- Mendel developed a method to "cross" (cross-pollinate) pea plants with contrasting traits.



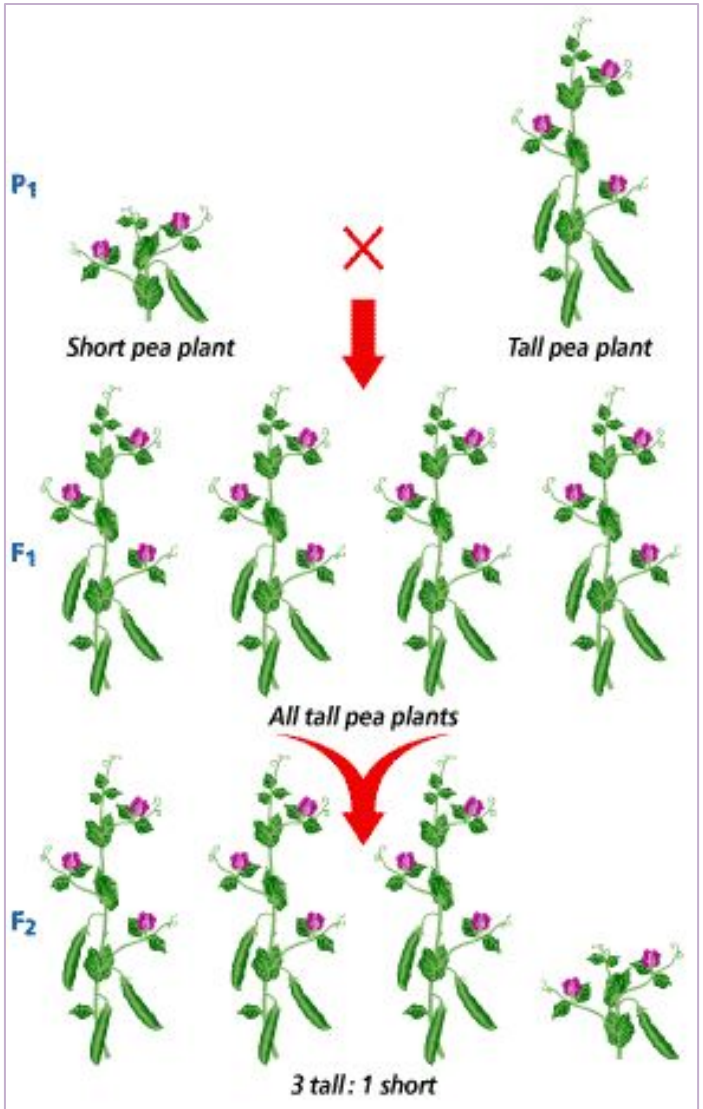


Mendel's Experiments – the first filial generation…

- First, he crossed purebred tall plants with purebred short plants to produce the first generation of offspring (today scientists call this the F1 generation).
- All of the offspring in this generation were tall.

Mendel's Experiments – the second filial generation…

- When the F1 generation was fully grown, he allowed them to self-pollinate.
- Surprisingly to Mendel, the plants in the F2 generation were a mix of tall and short plants.
- Mendel found that about three fourths of the plants were tall, while one fourth were short.



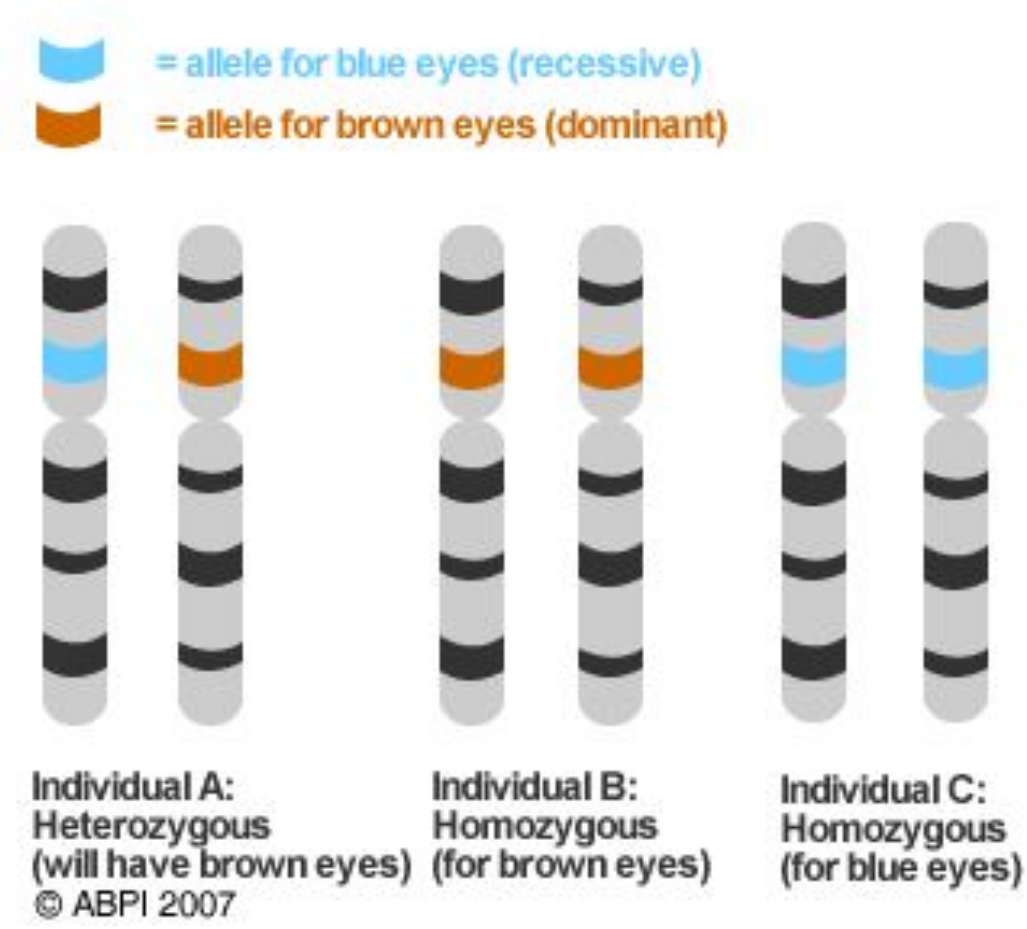
Mendel's Conclusions

- Mendel repeated his crosses for a variety of other traits and in every case only one form of the trait appeared in the F1 generation, but in the F2 generation the "lost" form of the trait always reappeared in about one fourth of the plants.

- Mendel came to some important conclusions:
  - Inheritance of each trait is controlled by "factors" (we now call these genes).
  - The factors that control each trait exist in pairs. For each trait, an offspring inherits one "factor" from each parent.
  - A trait may not show up in an individual but can still be passed on to the next generation.
  - One factor in a pair can mask another factor (i.e. the tallness factor masked the shortness factor in pea plants).
  - He realized that the principles of probability could be used to predict the results of genetic crosses.

Genes and Alleles

- Today we call the factors that control a trait genes. The different forms of a gene are called alleles.
- A dominant allele is one whose trait always shows up in the organism when the allele is present (i.e. tallness in pea plants)
- A recessive allele is hidden whenever the dominant allele is present. (i.e. shortness in pea plants)



Symbols for Alleles

- Geneticists use letters to symbolize alleles.
- Dominant alleles are represented by capital letters. For example, the allele for tall stems in pea plants can be represented as a **T**.
- Recessive alleles are represented by lower case letters. For example, the allele for short stems in pea plants can be represented as a **t**.
- Since offspring inherit two alleles for a trait (one from each parent), there are different possible combinations: **TT, Tt,** or **tt**.

Punnett Square

- A Punnett Square is a tool that shows all the possible genetic combinations that can result from a genetic cross. It can be used to determine the probability of a particular outcome.
- It was devised in the early 1900s by English geneticist Reginald C. Punnett.

Punnett's Squares

These show the 2 alleles of each parent plant crossed with each other and the resulting 4 possible offspring with T = tall, t = short. TT = dominant tall, tt = recessive short, Tt = mixed hybrid

TT = dominant tall (genotype tall, phenotype tall)
Tt = mixed hybrid (genotype hybrid, phenotype tall)
tt = recessive short (genotype short, phenotype short)

	T	T
T	TT	TT
T	TT	TT

Both parents are dominant tall so all offspring are tall.

	T	t
T	TT	Tt
t	Tt	tt

Both parents are mixed hybrids so offspring are a 3:1 ratio.

	T	T
T	TT	TT
t	Tt	Tt

One parent is dominant tall and one is mixed hybrid so all offspring are tall.

	t	t
t	tt	tt
t	tt	tt

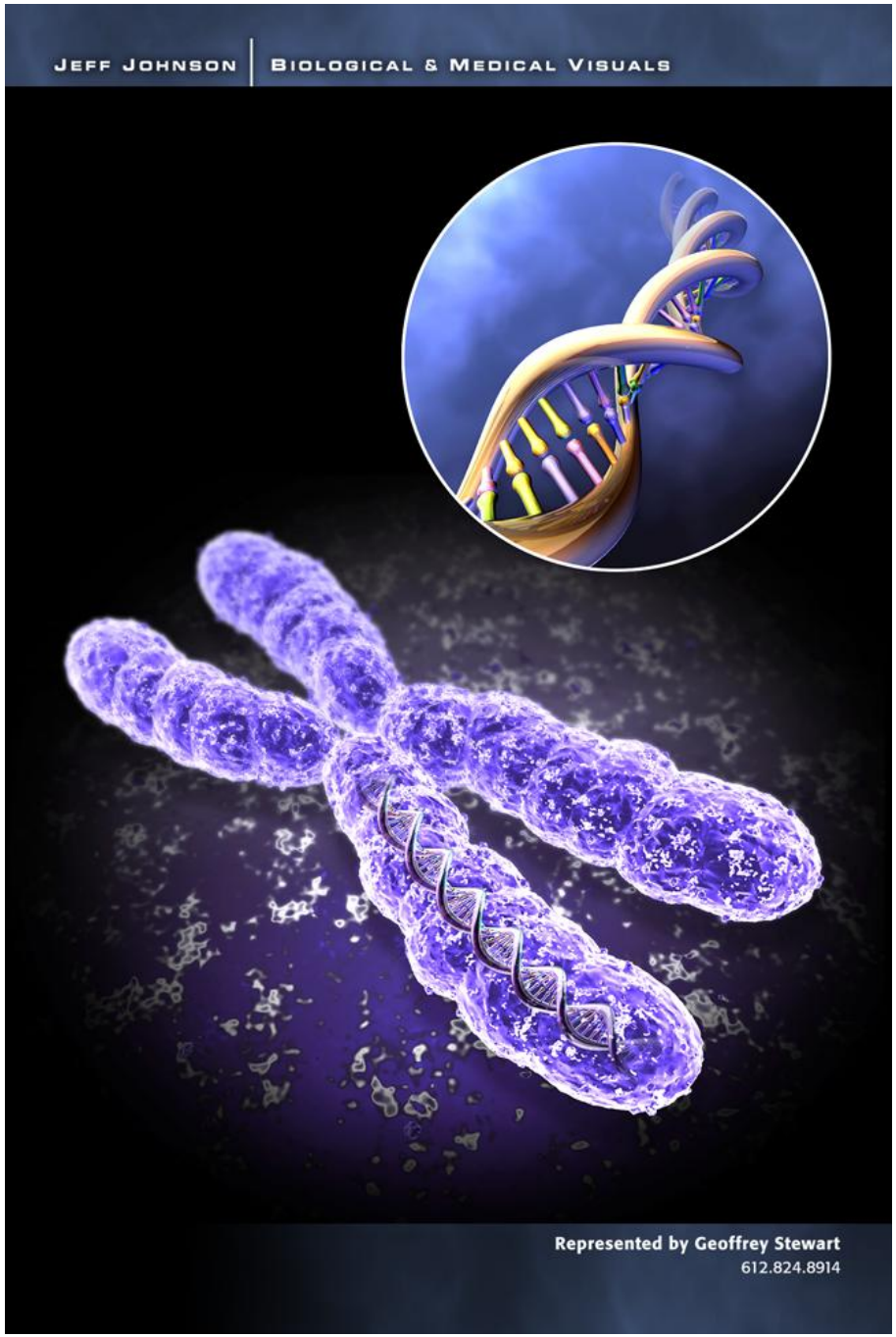
Both parents are recessive short so all offspring are short.

How Inheritance Works

- Mendel's work showed that genes existed, but scientists did not know where they were in cells until the early 20th century.
- In 1903, American geneticist Walter Sutton was studying grasshopper cells, paying attention to the movement of chromosomes during the formation of sex cells (i.e. sperm and egg). He believed that chromosomes were the key to understanding heredity.

Chromosomes

- A chromosome is a threadlike strand of DNA and protein found in cells that carries genetic information.



Chromosome Theory of Inheritance

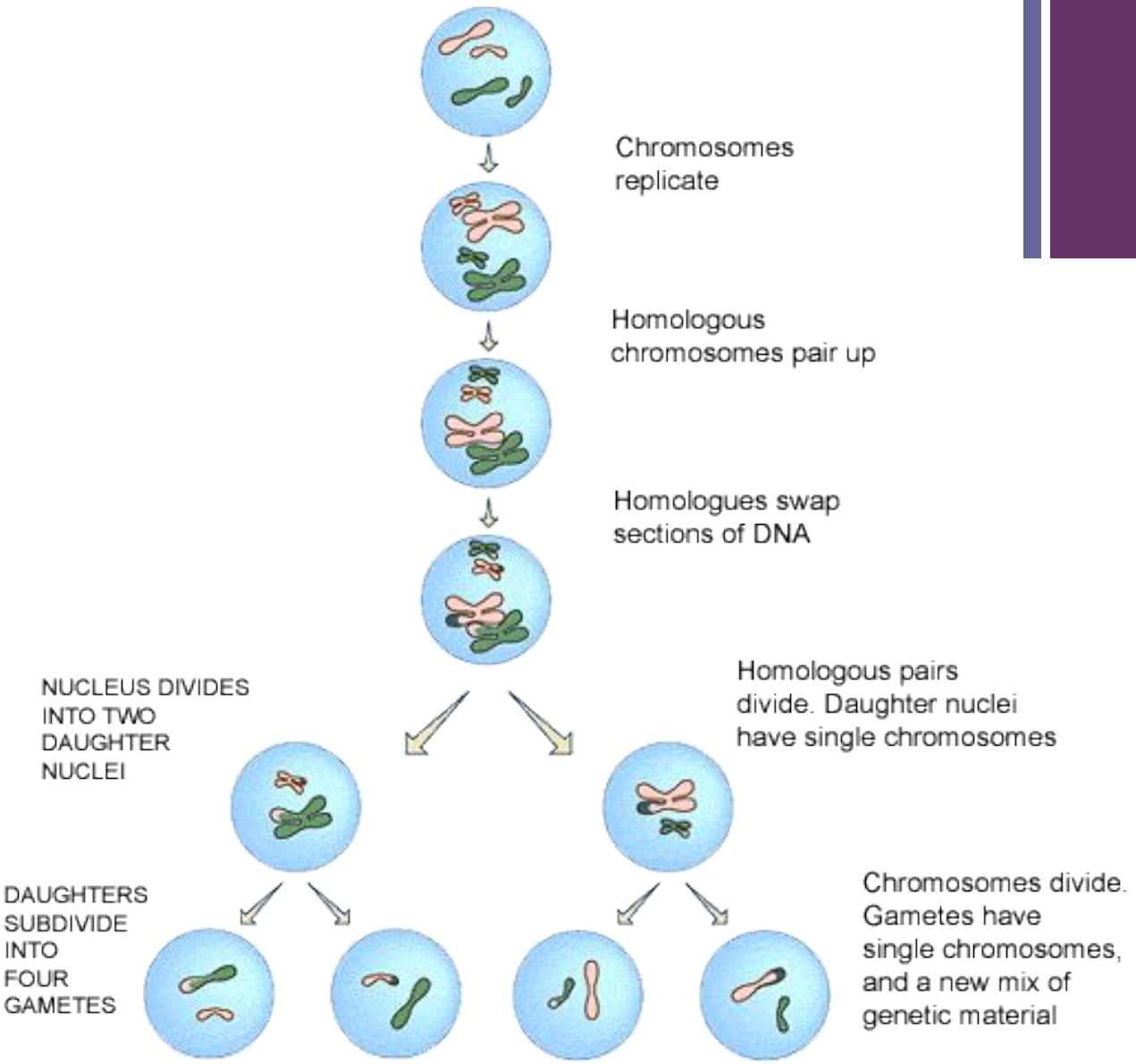
- Sutton found that the grasshopper's body cells had 24 chromosomes but that the sex cells had only 12; this led him to conclude that an organism's sex cells have exactly half the number of chromosomes found in the body cells.
- Sutton observed that during fertilization, a fertilized grasshopper egg again had 24 chromosomes.

Chromosome Theory of Inheritance

- Sutton found that the 24 chromosomes existed in 12 pairs—one chromosome in each pair came from the mother and one came from the father.
- From Mendel, Sutton knew that alleles existed in pairs (one from the mother and one from the father)—he concluded that the paired alleles were carried on paired chromosomes.
- This idea became known as the chromosome theory of inheritance, which basically states that **genes are carried from parents to their offspring on chromosomes**.

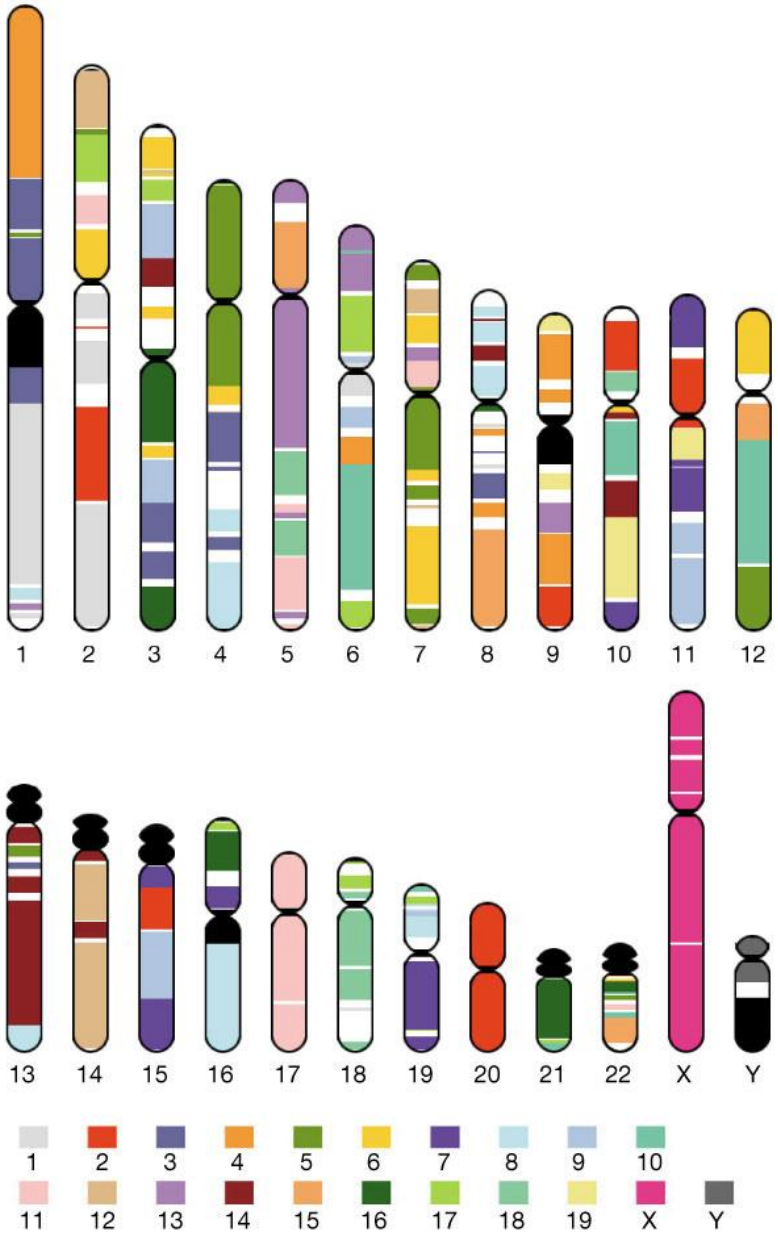
Meiosis

- The process by which the number of chromosomes is reduced by half to produce sex cells (sperm and egg) is called meiosis.



Genes

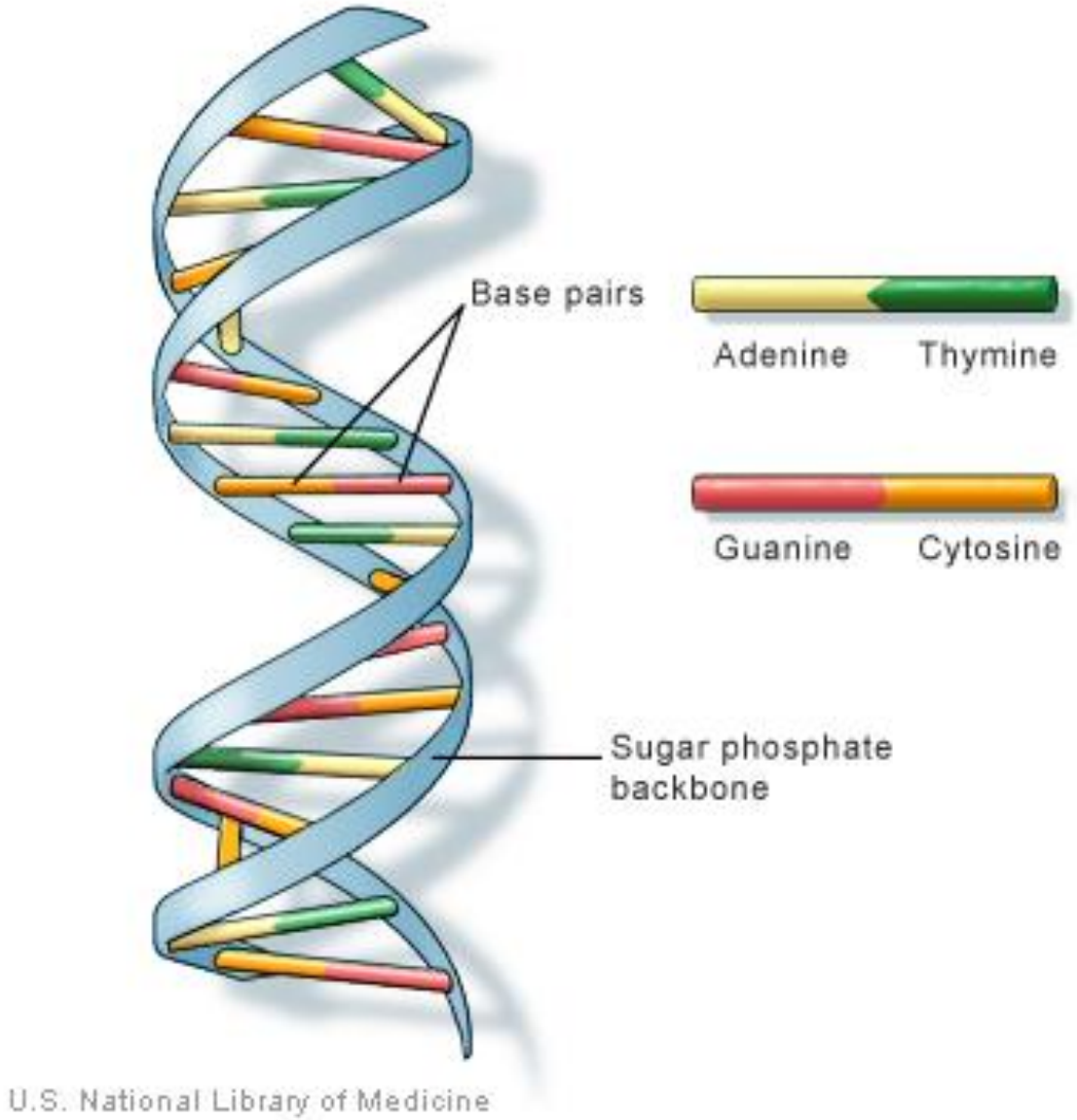
- Chromosomes are made up of many genes joined together.
- The body cells of human beings contain only 23 chromosome pairs (46 chromosomes) but between 20,000 and 25,000 genes, each of which controls a trait.



The Genetic Code - DNA

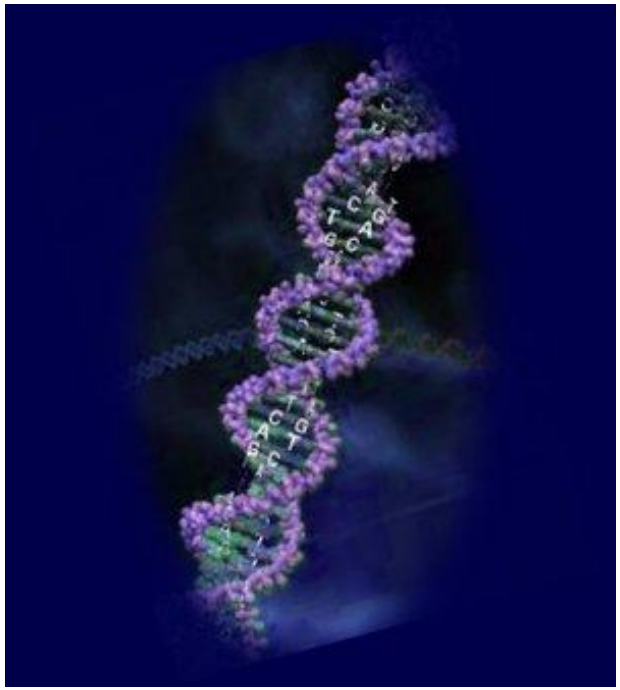
- The main function of genes is to control the production of proteins in organisms.
- Chromosomes are composed mostly of DNA (Deoxyribonucleic acid).
- DNA is made of four different nitrogen bases – adenine, thymine, guanine, and cytosine, which form the rungs of the DNA "ladder."
- A gene is a section of a DNA molecule that contains the information to code for one specific protein.

DNA


Base pairs
Adenine
Thymine
Guanine
Cytosine
Sugar phosphate backbone
U.S. National Library of Medicine

DNA

- The order of the nitrogen bases along a gene forms a genetic code that specifies what type of protein will be produced.
- The name of the "twisted ladder" shape of DNA is the **double helix**.



The Genetic Code - Mutations

- Any change in a gene or chromosome is called a mutation.
- Mutations can cause cells to produce an incorrect protein during protein synthesis and one result of this is that the organism's trait may be different from what it normally would have been.
- If a mutation occurs in a body cell, it will not be passed on to offspring; but if it occurs in a sex cell, the mutation could be passed on to offspring.
- Mutations might occur if one base is substituted for another, if one or more bases are removed from a section of DNA, or if chromosomes don't separate correctly during meiosis.

Mutations

- Mutations introduce changes in an organism and are therefore a source of genetic variety.
- Some mutations are harmful, a few are helpful, and some are neither.