

# Topic 1 – Genes

# Genes

Inherited traits:

- Color blindness (on X chromosome)
- Cleft chin – may be inherited

Not simple inheritance:

- Freckles
- Ability to taste Phenylthiocarbamide (in cabbage, brussel sprouts, broccoli)

Humans have about 20,500 genes

Most variations in genes are harmless but some cause serious illnesses or disabilities – a driving force for studying genetics

# Genes and Gene Sequences

## Cystic Fibrosis

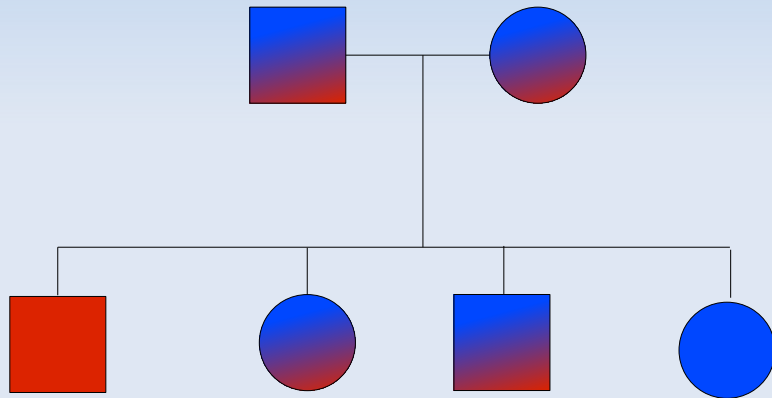
- A "rare" condition, the most widespread life-shortening genetic disease
- 30,000 people in US have it
- 1 in 30 Caucasian Americans is a carrier
- 1 in 46 Hispanics is a carrier
- 1 in 65 African Americans is a carrier
- 1 in 80 Finns
- 1 in 90 Asians

No cure. Symptoms: intestinal obstruction, infertility, life-threatening clogging and infection of lungs.

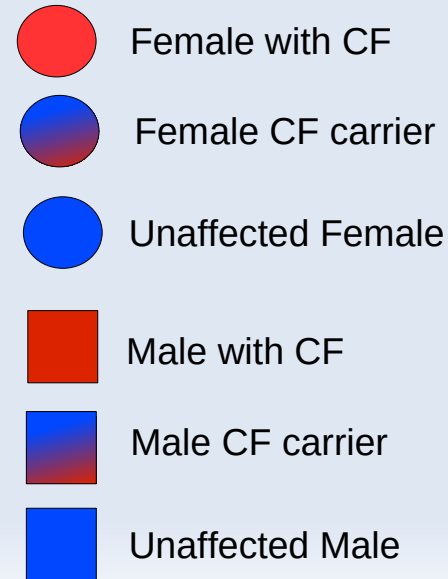
Before 1939: most CF patients died before 2<sup>nd</sup> year of life. It's better now.

# Genes and Gene Sequences

A possible *Pedigree chart* for Cystic Fibrosis carriers:



If two CF carriers have children,  
there is a 1 in 4 chance a child  
will have CF

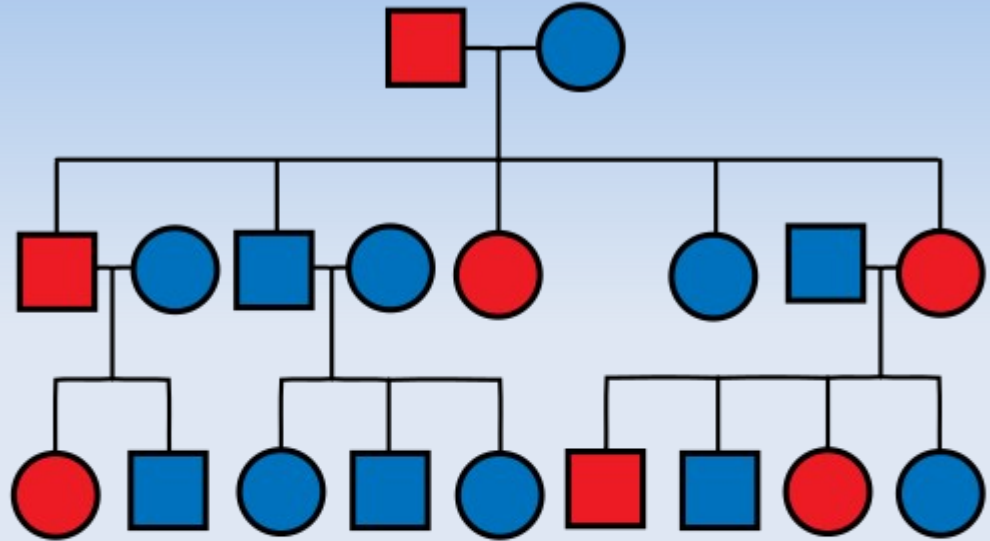


Cystic Fibrosis is Autosomal recessive  
A carrier may have no symptoms b/c they will have two copies of the gene and if one is  
"healthy", the individual will be healthy.

# Genes and Gene Sequences

A different pedigree chart

Huntington's disease  
Autosomal dominant



KEY



Affected  
Male



Affected  
Female



Unaffected  
Male

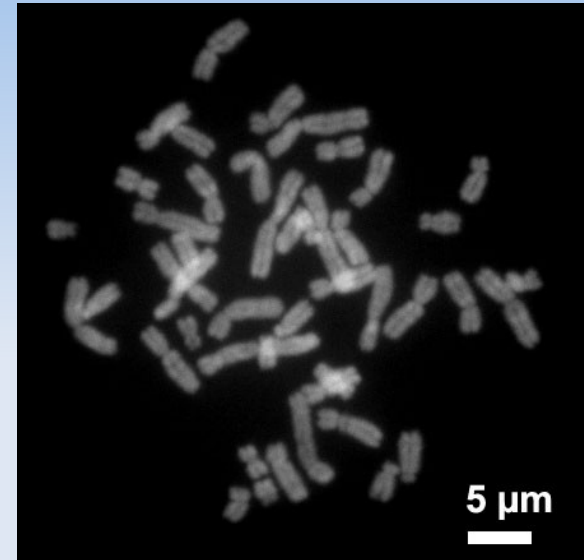
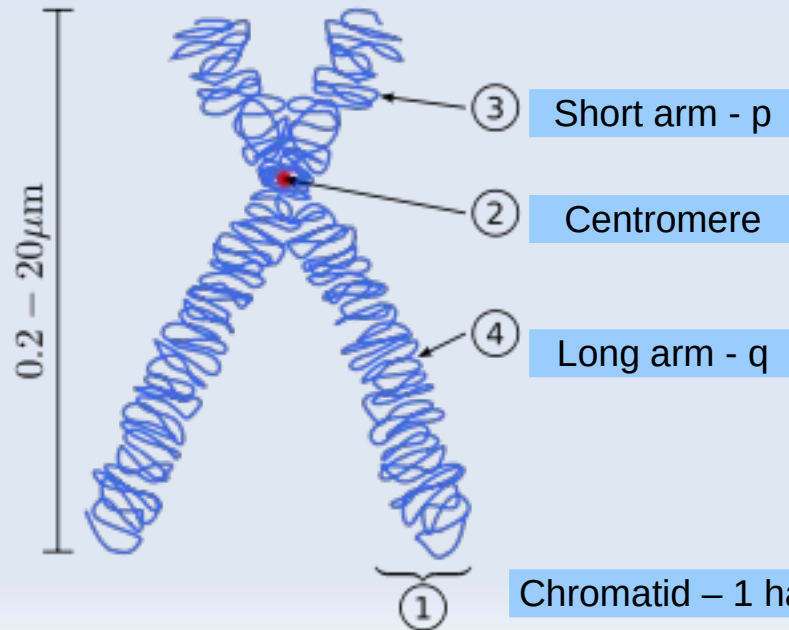


Unaffected  
Female

# Genes and Gene Sequences

An individual's inherited characteristics are transmitted via chromosomes

We have 23 pairs: 22 are not involved in determining sex (autosomes) and have an X shape.

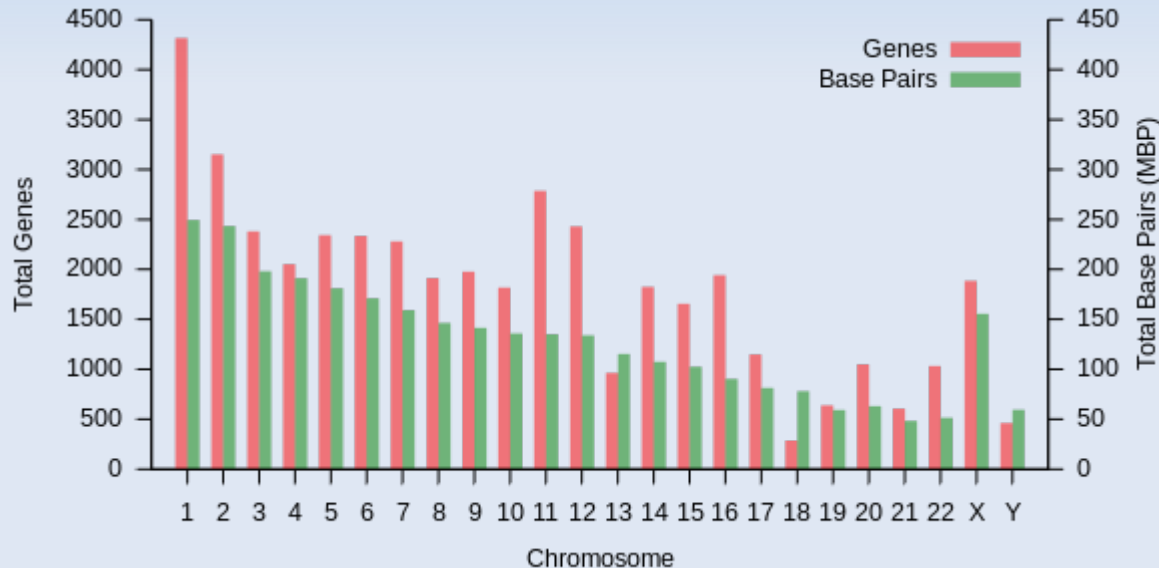


Wikipedia

# Genes and Gene Sequences

The 23 pairs have different sizes and different numbers of genes – the autosomes are numbered in order of decreasing size.

When unwound, chromosome 1 is 15 cm ( $1\frac{1}{2}$ ') long.



Wikipedia

The "**locus**" of Cystic Fibrosis is 7q31.2 – long arm of the 7<sup>th</sup>.

# Genes and Cells

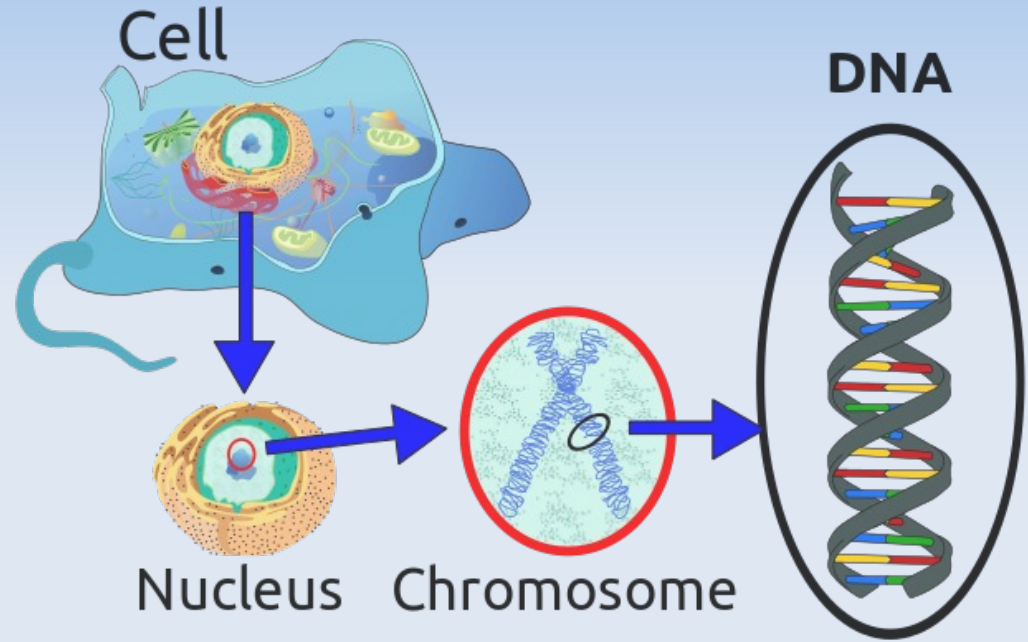
Every cell in our body except for mature red blood cells has DNA.

Most of these cells have a full set of 46 chromosomes

The two exceptions are egg cells in women and sperm cells in men – they have 23 chromosomes each.

We get a complete copy of a genome (~20,500 genes) from each parent.

Wikipedia



When unwound, the DNA in a single cell will stretch out to 6 feet.

To see how DNA fits in with the rest of our body, watch this NOVA animation called [Journey into DNA](#)

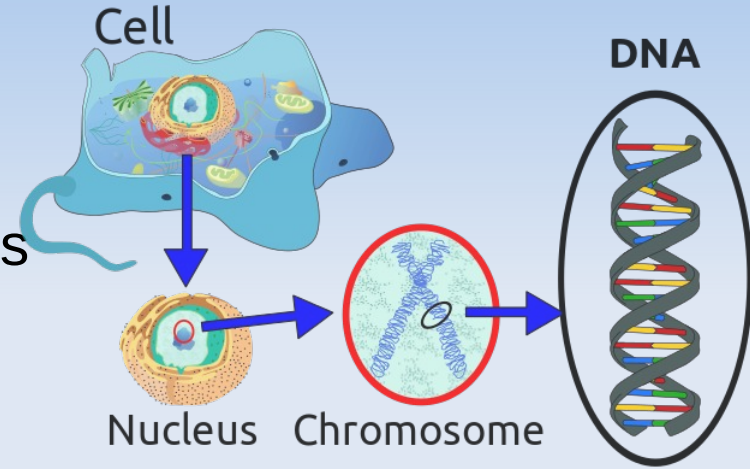


# Genes and cell function

One gene makes one protein or RNA product.

In 1989, it was determined that the Cystic Fibrosis gene codes for a protein molecule called the **Cystic Fibrosis Transmembrane conductance Regulator (CFTR)**

CFTR controls the the production of sweat, mucus, and digestive fluids.

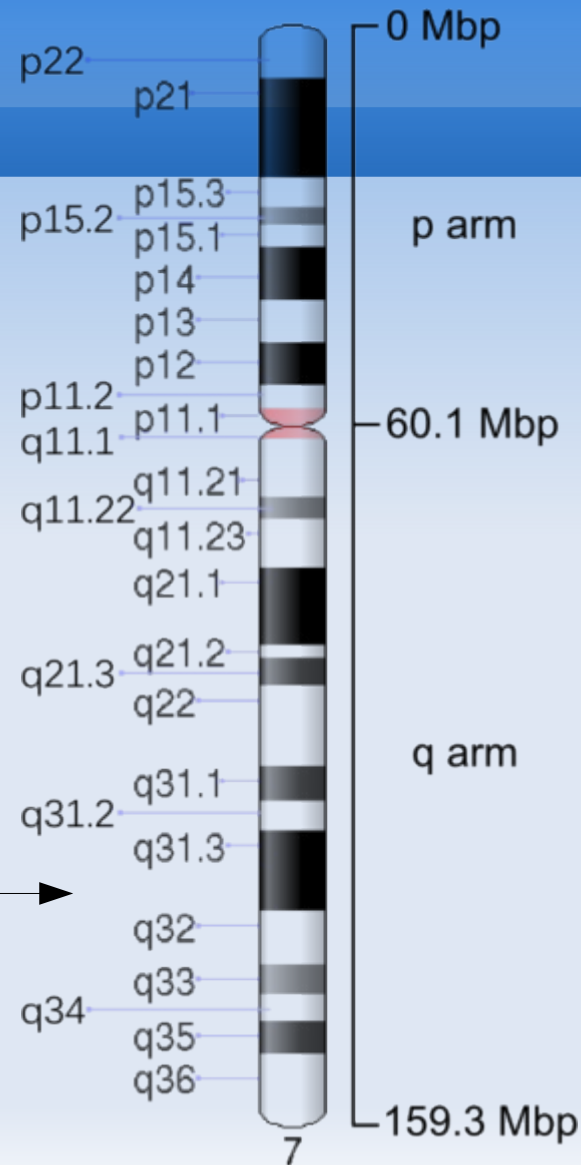


# CFTR

CF carriers have one functional and one defective gene. The functional gene on one chromatid can compensate for the defective one in the same location on the other chromatid.

If both copies are defective, the individual will have defective CFTR proteins that are unable to regulate transport of chloride ions → lungs are clogged with mucus → infections.

Locus of  
CFTR gene

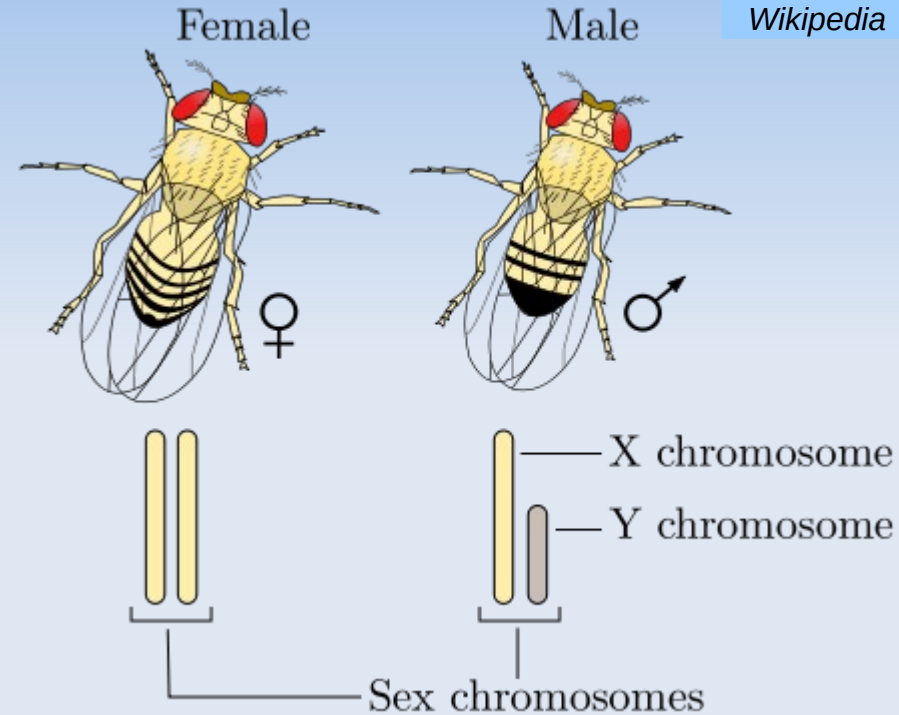


# XY sex-determination

Female humans have two X chromosomes.

Male humans have an X and a Y chromosome with 600 fewer genes than the X

Intersex people can have various combinations: XXY, XYY, XXYY



*Drosophila* have XY sex-determination

Some animals (chickens, butterflies, Komodo dragons, etc.) have ZW chromosomes

Plants have more complex genes for sex determination;  
Many plants are monoecious - e.g most roses have both sexes in each flower; mostly diploid or tetraploid, but also triploid, pentaploid

# Color Blindness and XY chromosomes

There are many kinds of color blindness – not all are inherited.

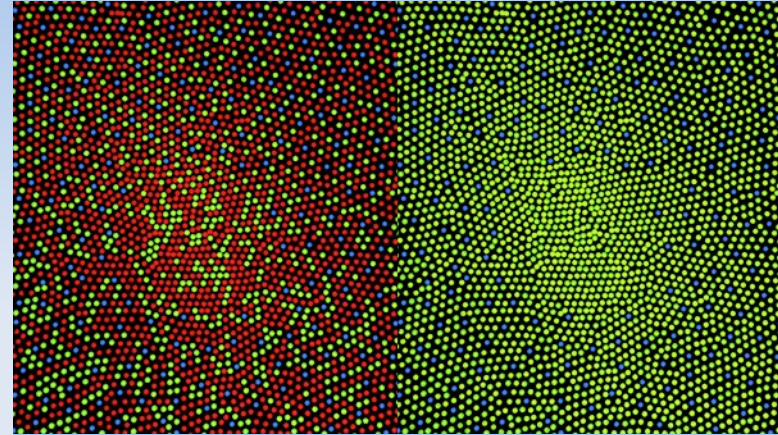
Color blindness due to absence or mutation of the red or green retinal photoreceptors is X-linked.

Genes for red and green color receptors are on the X chromosome.

Males are color blind if their single X chromosome has a mutation.

Females are color blind if both X chromosomes have the mutation.

Wikipedia



Distribution of cone cells in normal (left – RGB) and color blind (right – GB) individuals

Gene for blue receptor is on chromosome 7 – not sex-linked. Males and females are equally susceptible.

# DNA research: US Dept. of Energy

genomics.energy.gov

This site is a gateway to the U.S. Department of Energy's genomic websites



With the development of the atom bomb, the dangers of radiation to human health came to be well recognized.

1946 Nobel prize in Physiology went to HJ Muller for effects of radiation

1947: Atomic Bomb Casualty Commission (funded by Atomic Energy Commission, part of DOE in 1970) funded research on genetics: mechanisms of mutagenesis and effect of radiation on genes.

The DOE funded genomics research leading to the largest biological research project ever, the 13-year **Human Genome Project**.

Still funds a lot of biology & environmental projects.

# Similarities and Variations

Many mammals – mice, rats, humans, chimpanzees, etc. - have about 20-25,000 genes.

Big surprise when we found out: our genome is not much different from mice!

If you were to be able to compare your genome with that of someone (of the same gender) sitting next to you, you will find a 0.1% difference – 3 million differences in 3 billion nucleotides – i.e. 1 difference every 1000 bases.

The difference with chimpanzee genome: every 100 bases – 1% difference.

A single letter difference can lead to a mutated gene resulting an inherited illness or an enhanced physical ability

**So, what exactly is a "gene"?**

# So, what exactly is a "gene"?

Simple question.

If only we had a simple answer!

One answer: "The basic physical and functional unit of heredity"

Or try: <http://learn.genetics.utah.edu/content/molecules/gene/>

Or this: <https://www.youtube.com/watch?v=5MQdXjRPHmQ>

The <https://en.wikipedia.org/wiki/Gene> Wikipedia page may help.