BB 101

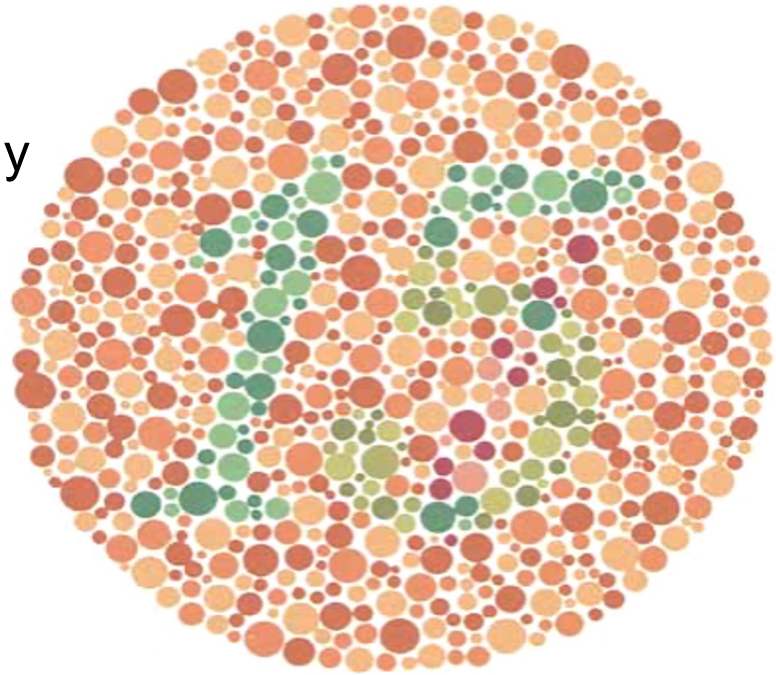
Genetics & DNA as a genetic material

Tutorial 3

25.01.2024

Examples of Sex-linked Traits And Disorders

- Red-green Colour blindness
- Duchenne muscular dystrophy
- Night blindness
- Hemophilia



Transmission of X-linked Recessive Traits: Red-green Color Blindness

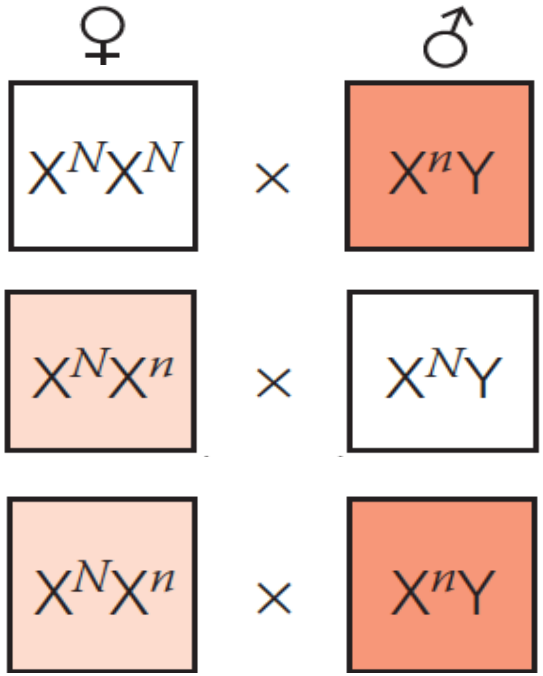
Red-green Color Blindness: X-linked Disorder: A re-cap!

***N** = dominant allele for normal color vision (carried on X chromosome)*
***n** = recessive allele having a mutation for color blindness*

Unaffected individuals

Carriers

Color-blind individuals



Fathers pass X-linked alleles to all of their daughters but to none of their sons.
Mothers can pass X-linked alleles to both sons and daughters.

Any male receiving the recessive allele from his mother will express the trait.
Therefore, far more males than females have X-linked recessive disorders.

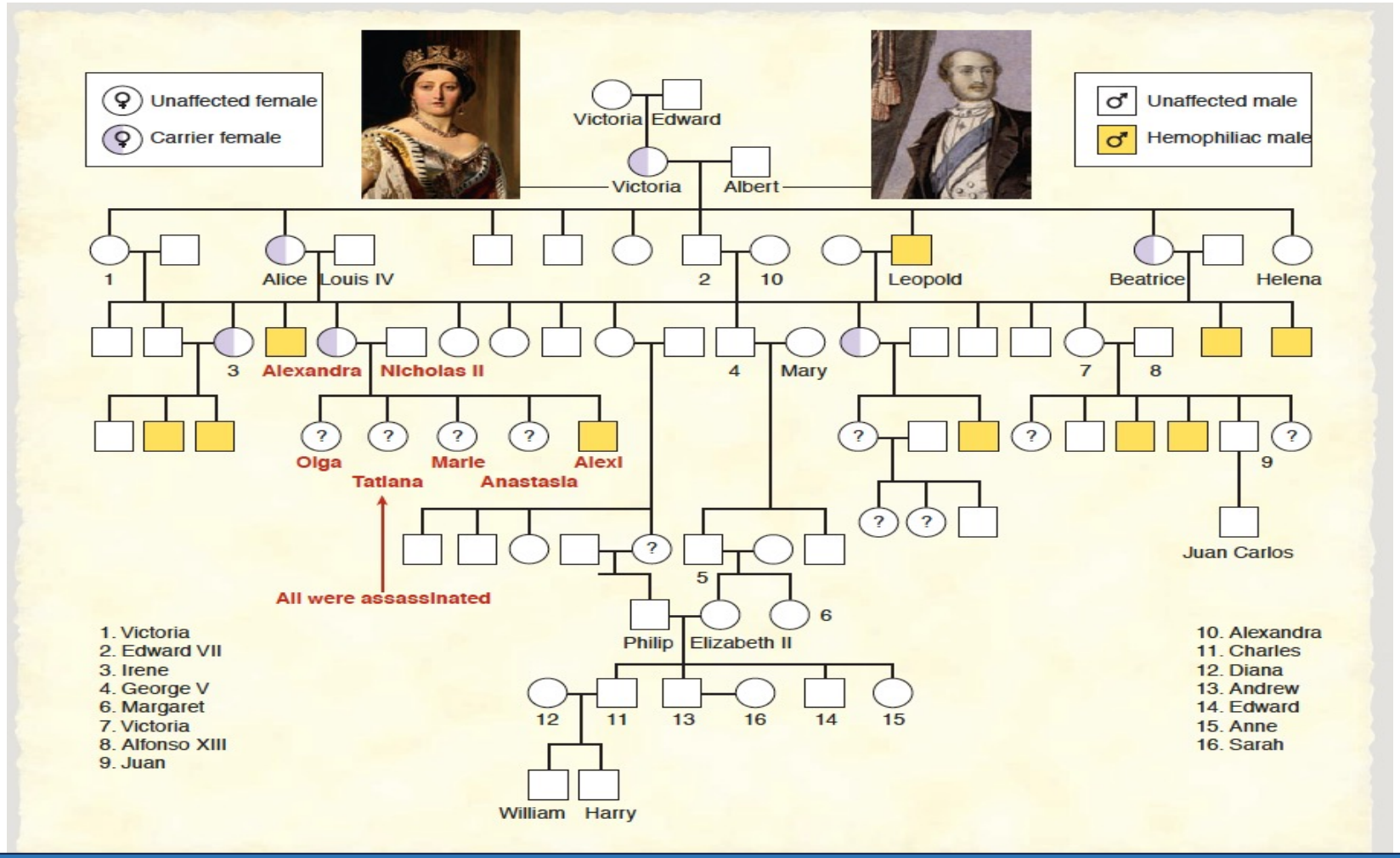
X-linked Recessive Disorder: Hemophilia

- Absence of one or more of the proteins required for blood clotting.
- When a person with hemophilia is injured, bleeding is prolonged because a firm clot is slow to form.
- Small cuts in the skin are usually not a problem, but bleeding in the muscles or joints can be painful and can lead to serious damage

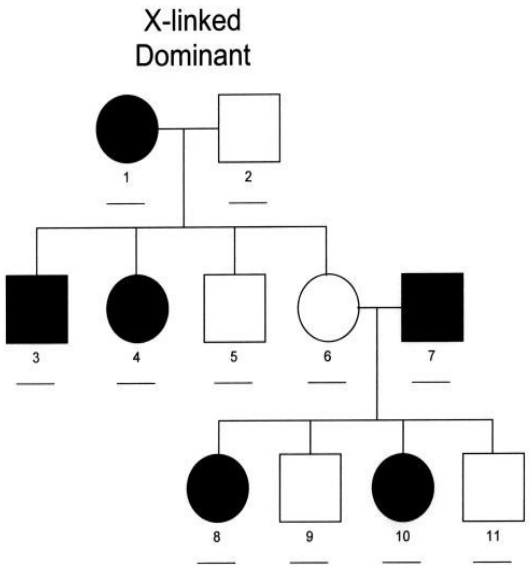
Hemophilia and Royal Family of Europe

- Queen Victoria of England is known to have passed the allele to several of her descendants.
- Intermarriage with royal family members of other nations, such as Spain and Russia, further spread this X-linked trait.

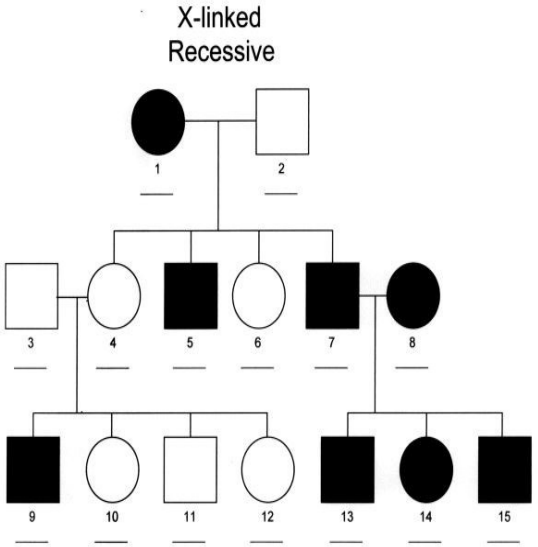
Example: Royal Families of Europe And Hemophilia



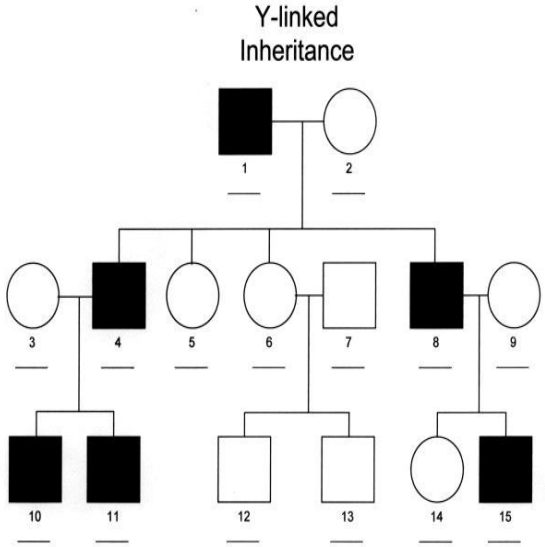
PEDIGREE ANALYSIS OF SEX LINKED DISORDERS



X-linked dominant



X-linked recessive



Y-linked

The genetic basis of the mutation, and how it resulted in a nonfunctional blood-clotting factor, is now understood..

Today, people with hemophilia are treated as needed with intravenous injections of the protein that is missing

PROBLEMS OF GENETICS

In mice, black coat color (*B*) is dominant over brown (*b*), and a solid pattern (*S*) is dominant over white spotted (*s*). Color and spotting are controlled by genes that assort independently. A homozygous black, spotted mouse is crossed with a homozygous brown, solid mouse. All the F1 mice are black and solid. A testcross is then carried out by mating the F1 mice with brown, spotted mice.

a. Give the genotypes of the parents and the F1 mice.

b. Give the genotypes and phenotypes, along with their expected ratios, of the progeny expected from the testcross.

Ans: a. Parents – BBss, bbSS; F1 – BbSs

b. Genotype – Bbss, BbSs, bbss, bbSs; Ratio – 1:1:1:1

Phenotype – Black Spotted, Black Solid, Brown Spotted, Brown Solid

PROBLEMS OF GENETICS

Haemophilia (reduced blood clotting) is an X-linked recessive disease in humans. A woman with haemophilia mates with a man who exhibits normal blood clotting. What is the probability that their child will have haemophilia?

Ans: ½. All the male progenies will be affected.

Why should we know about genetic material?

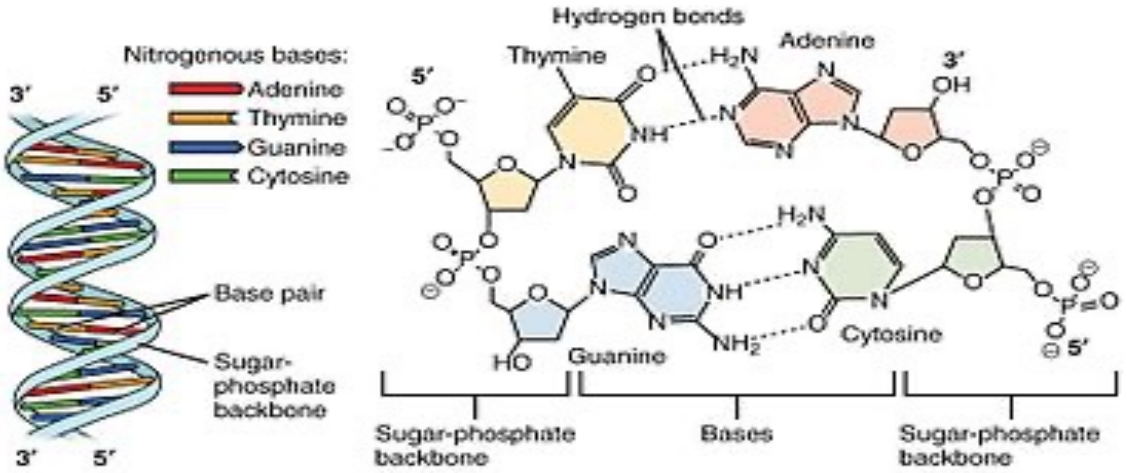
1. The Blueprint of life,
2. Pass genetic information from one generation to the next.
3. Diagnosing genetic disorders, predicting susceptibility to diseases, and developing personalised medicine.

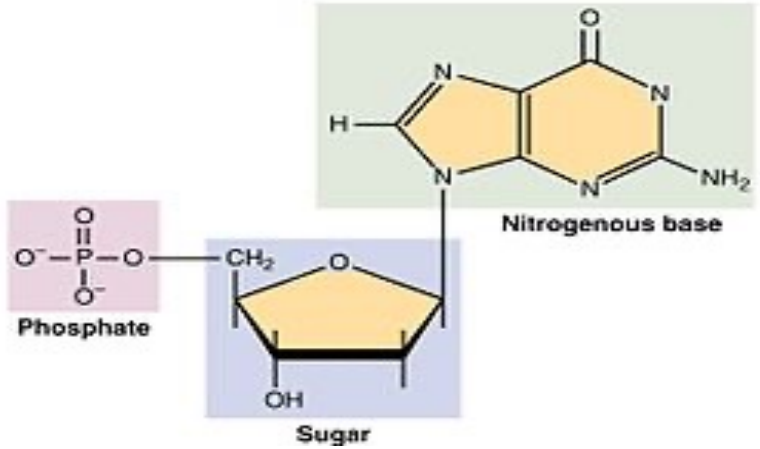
- But which of them is the genetic material and how scientists concluded?

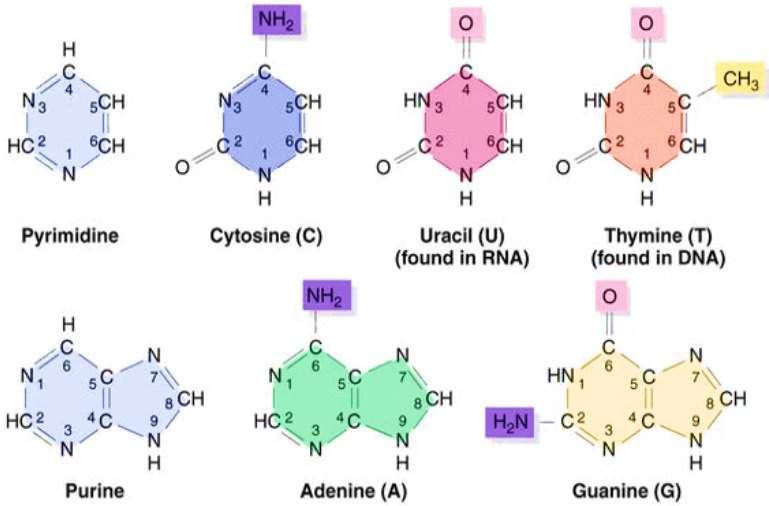
https://as1.ftcdn.net/v2/jpg/05/41/59/98/1000_F_541599815_0DeKJUZLUCdvIBtpAQ955DnbBtoOcbs5.jpg

https://www.ck12.org/flx/show/image/201301131358096717284223_df0d2514c98b5080c8eedc3e48ee5a55-201301131358098216540895.jpg

COMPOSITION OF DNA







Notice the directionality of both strands!

https://www.genome.gov/genetics-glossary/Deoxyribonucleic-Acid

ELUCIDATING THE DNA STRUCTURE

What was already known: DNA is a polymer consisting of A, C, G and T (referred to as nucleotide bases)
(This was based on Fedrick Miescher Albrecht Kossel's research)

CHARGAFF'S OBSERVATIONS:

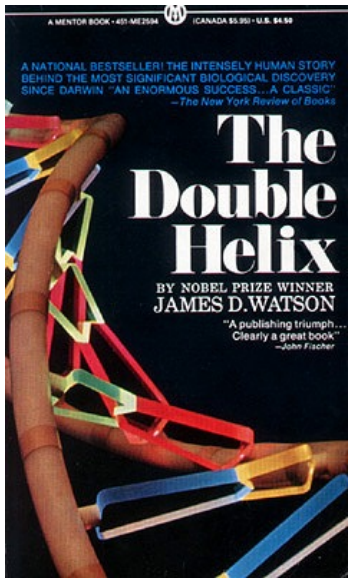
Observation 1: Chargaff noted that the nucleotide composition of DNA varies among species

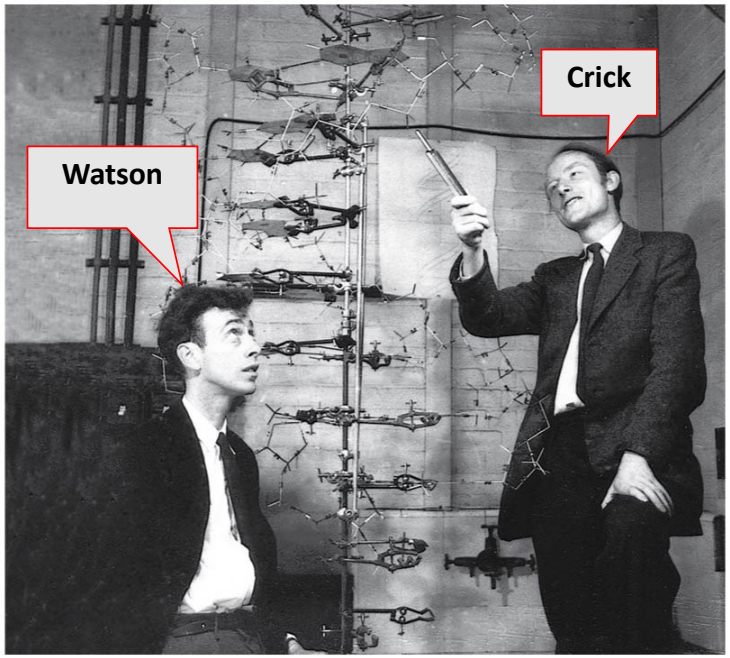
What does it actually imply?

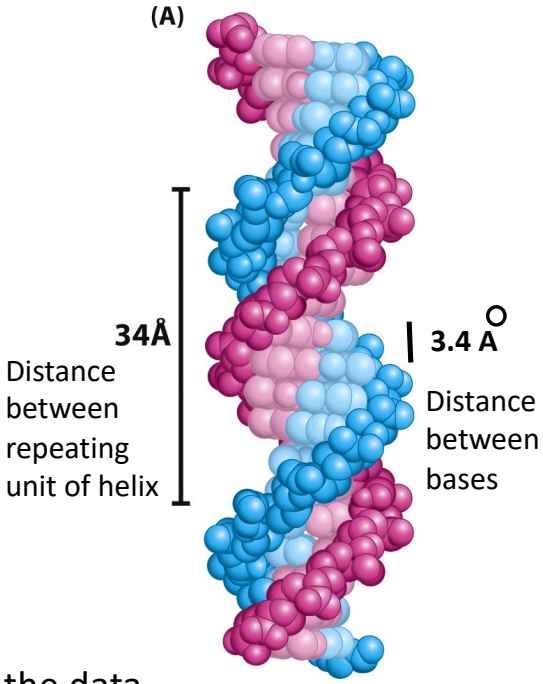
Observation 2: No. of A ≃ No. of T; No. of G ≃ No. of C

DISCOVERY OF STRUCTURE OF THE DNA

Watson-Crick Model of Double-Helical DNA



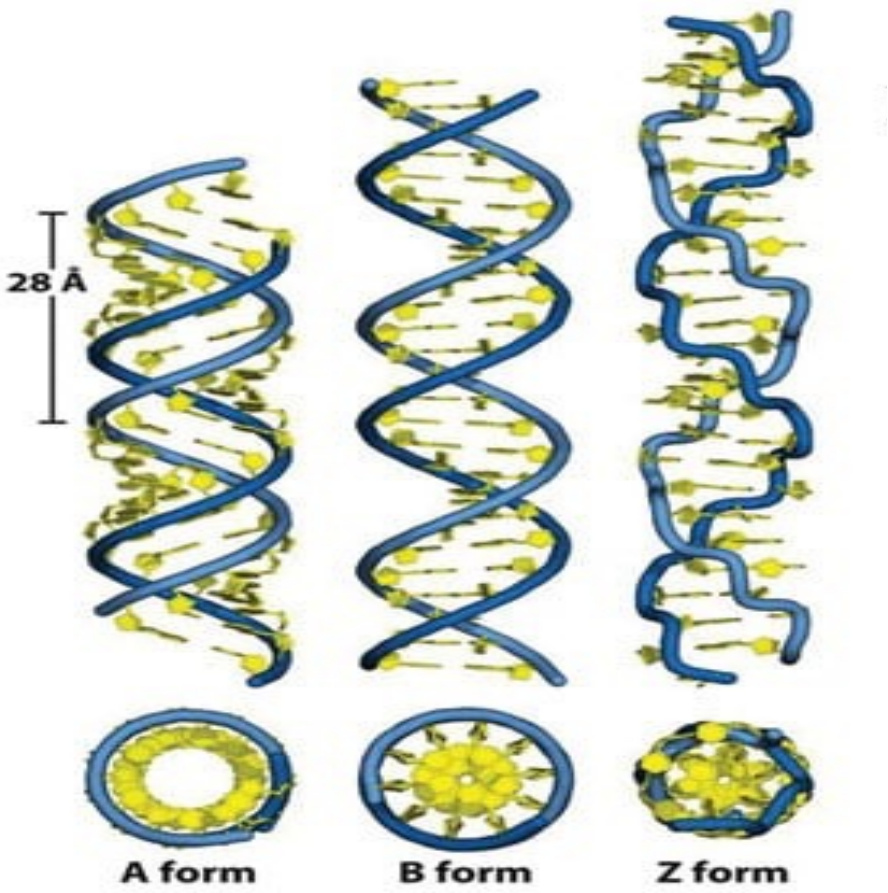




Watson and Crick played with chemical models to come up with a structure that matched the data

BUT DOUBLE HELIX IS NOT THE ONLY STRUCTURE OF DNA…

https://www.nature.com/scitable/topicpage/discovery-of-dna-structure-and-function-watson-397/



https://microbenotes.com/different-forms-of-dna-b-form-a-form-z-form/

STRUCTURE AND COMPOSITION OF DNA

1. If we find the composition of 'C' in DNA to be 20%, what do you think is the composition of base 'A'?

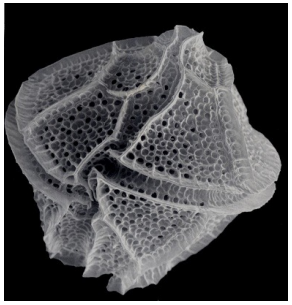
Ans. 30%

1. If the sequence of DNA in the parent stand is 'AGTCC', what do you think is the sequence in the daughter strand strand?

Ans. TCAGG

Does genome size correlate with “complexity”?

Human
3 billion
bp (base
pairs)



An alga
~98 billion bp

Onion
~16
billion bp

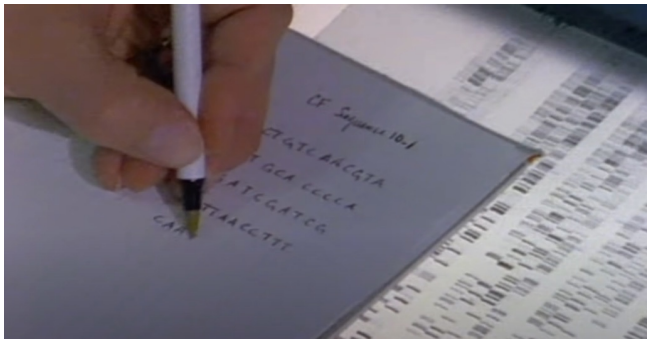
Marbled
lungfish
~130
billion bp

The length of total DNA in our body??

https://humgenomics.biomedcentral.com/articles/10.1186/s40246-022-00396-x

HUMAN GENOME PROJECT

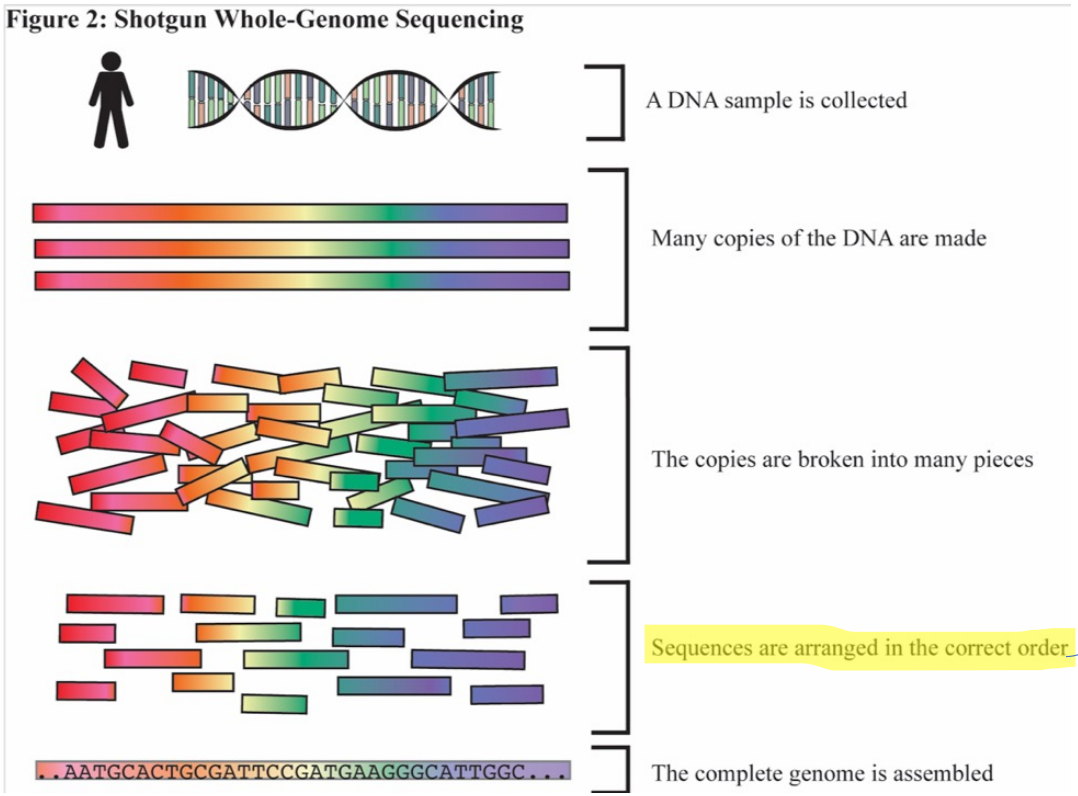
Size of human genome: 3.2 billion base pairs

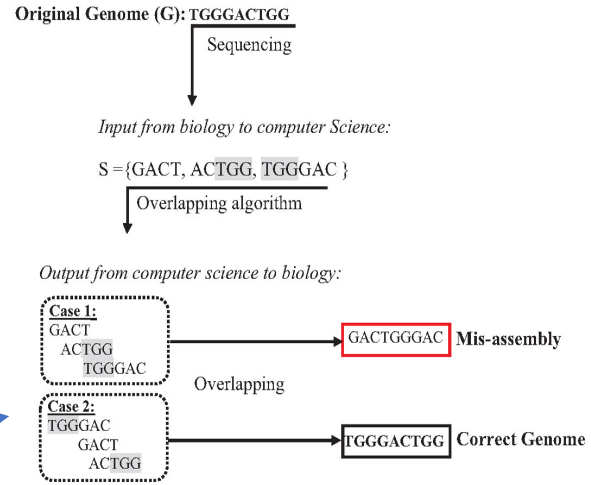


Estimated time for sequencing human genome **BY HAND**: at least 100 years

Computational technologies entered HGP by late 1990s, and the project was finished by 2003

HUMAN GENOME PROJECT – Sequencing the 3 billion bp human genome





YouTube video on HGP: https://youtu.be/qOW5e4BgEa4?si=xsNUC8N-alGADMtl

Application of genome sequencing

- Single base changes between reference and patient genomes

Disease Susceptibility, Diagnosis, Personalized Medicine