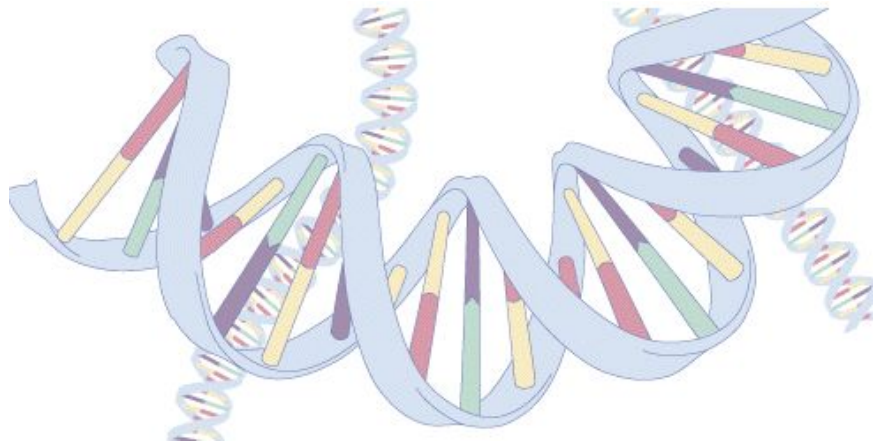


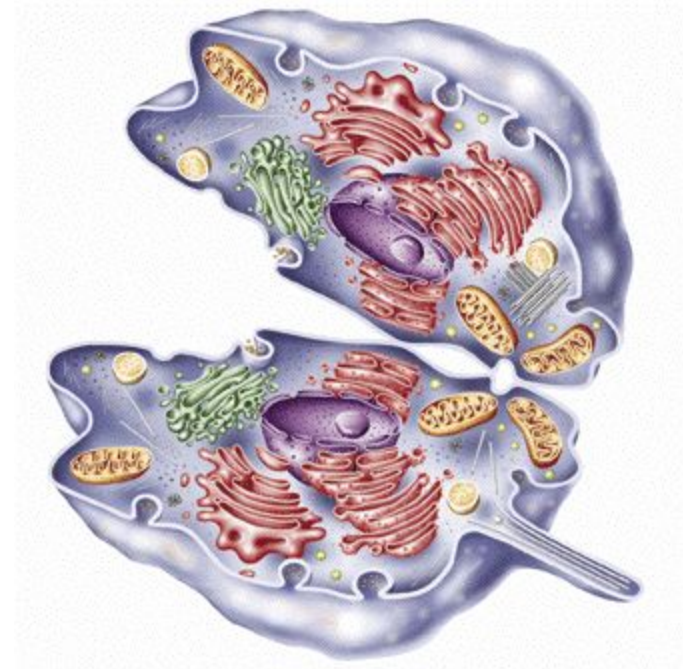


DNA, Genes and Genetics

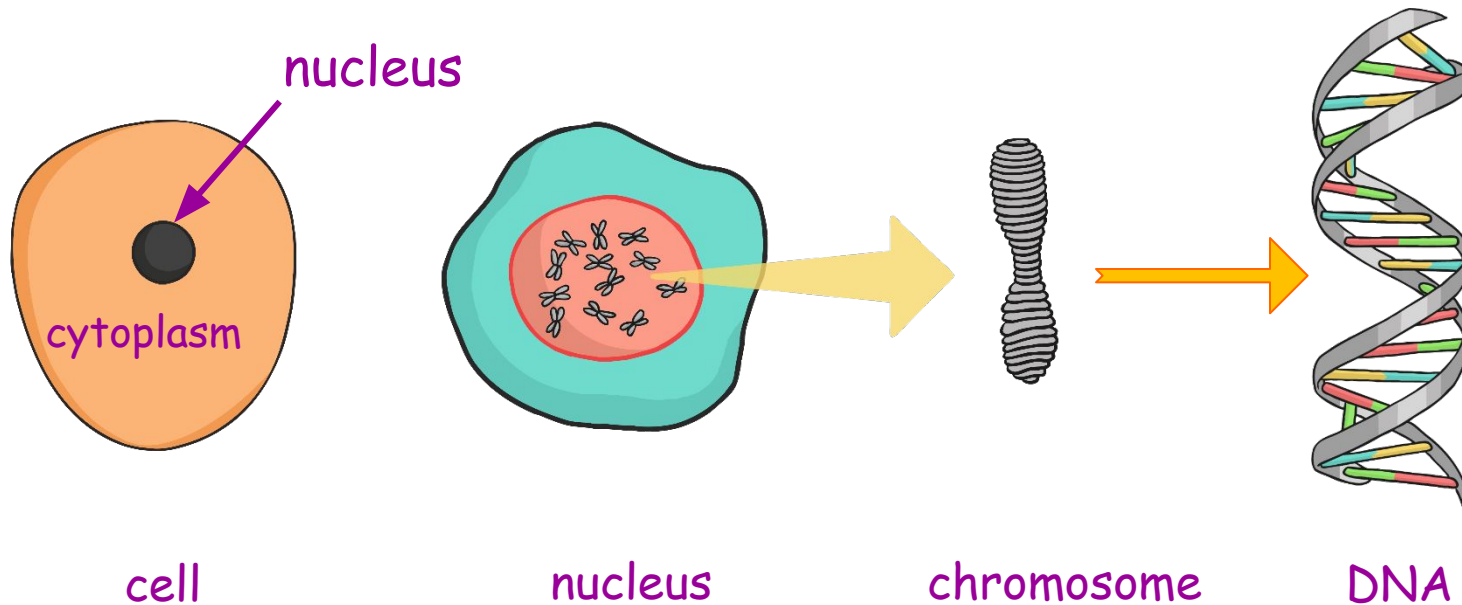
John Strouboulis
King's College London
john.strouboulis@kcl.ac.uk

The Cell

- Basic component of life
- All cells share basic organisation and structure, e.g. nucleus, mitochondria etc.
- They are highly specialised depending on the tissue they're in and functions they execute

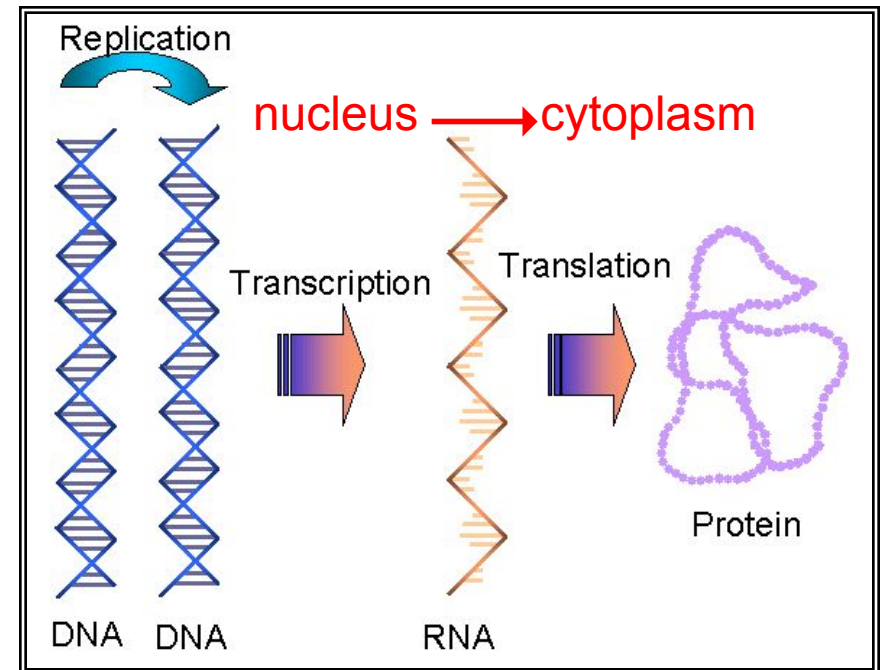


- Genetic material is located in nucleus
- The genetic information is stored in Deoxyribonucleic acid, DNA
- DNA contains the information needed to build an individual



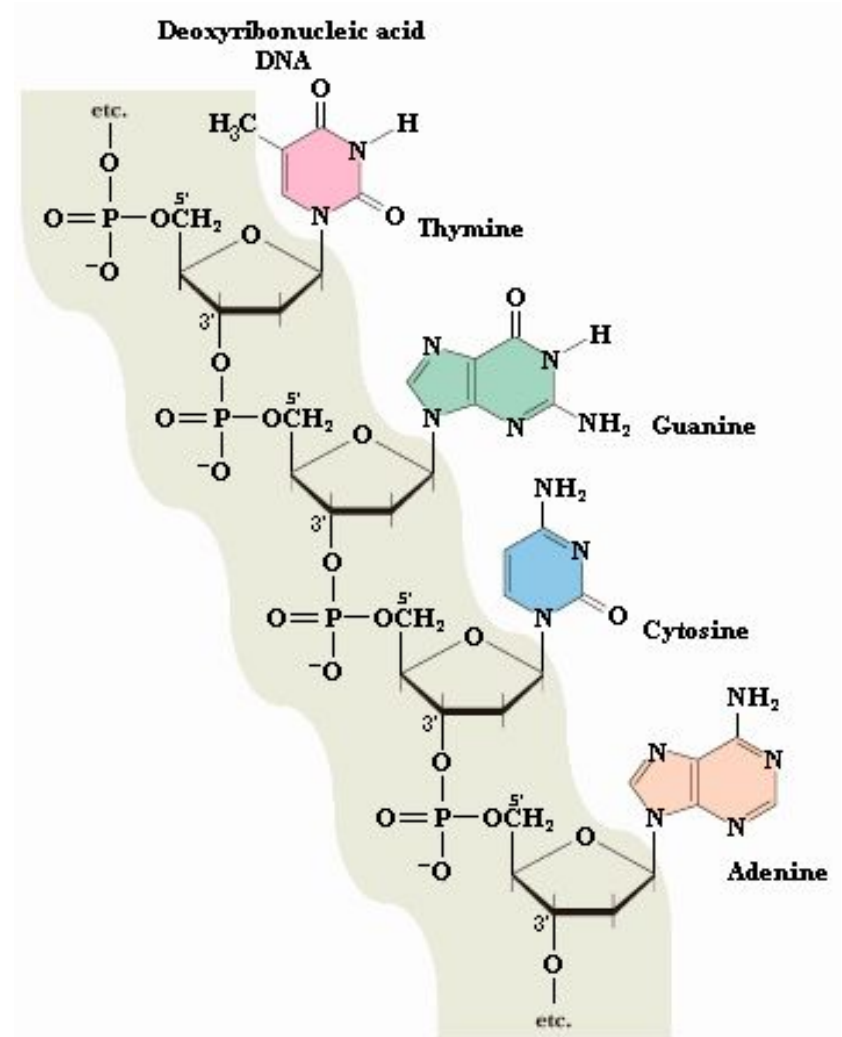
What is DNA needed for?

- Genetic information stored in the DNA is copied and converted to protein
- RNA molecules work as messengers
- Proteins are the biological workers

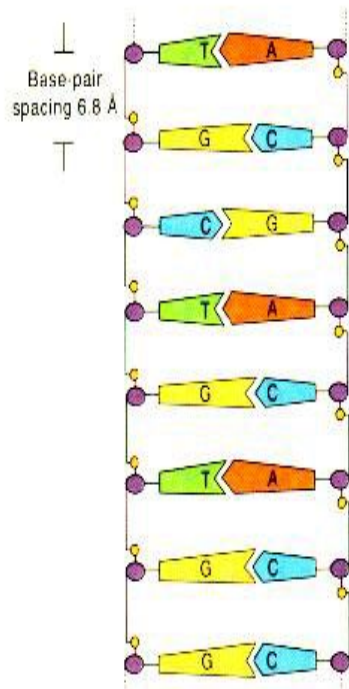


What is DNA?

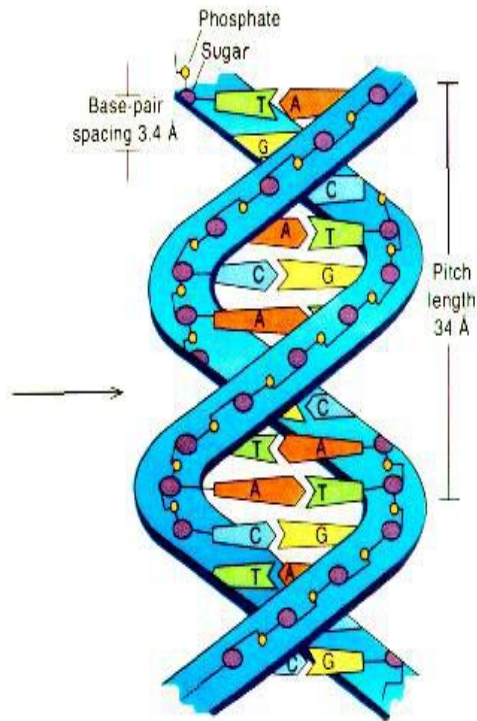
- DNA is made up of 4 building blocks called **nucleotides**:
 - Adenine (A)
 - Cytosine (C)
 - Guanine (G)
 - Thymine (T)
- They join up to form **VERY** long chains



The DNA double helix



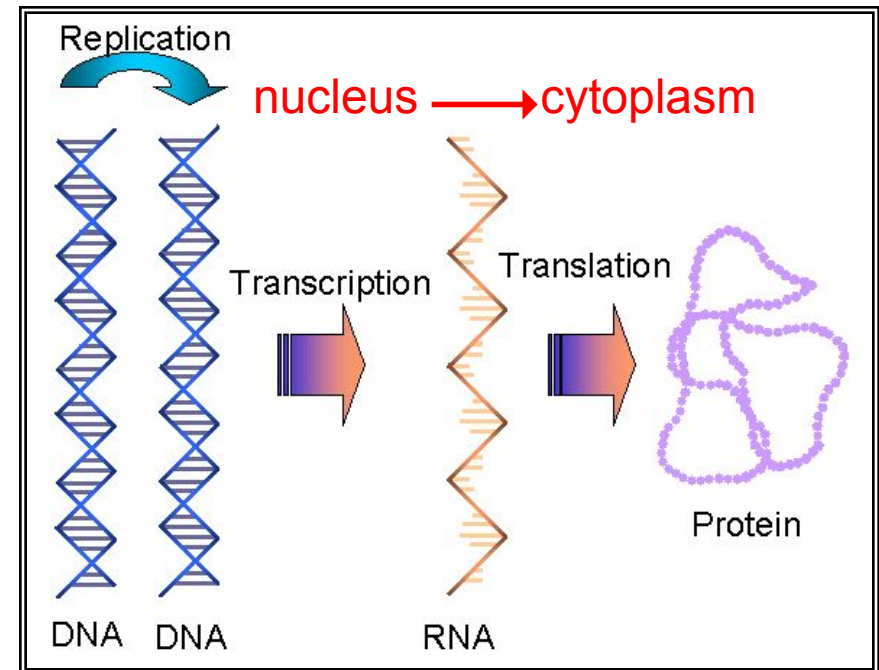
"ladder"



Double helix

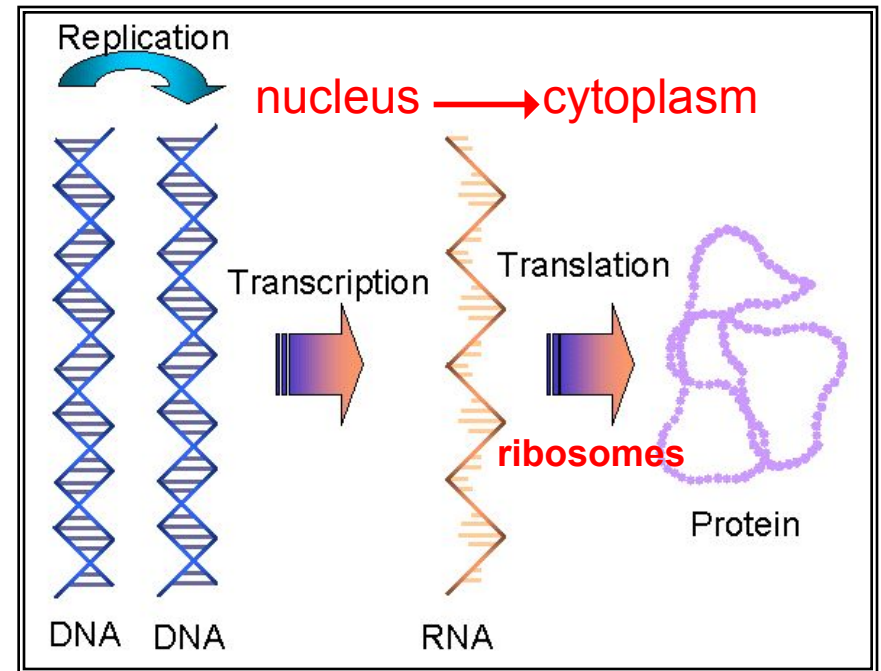
- Two chains are joined and twisted to form the double helix!
- The pairing up of the building blocks (**nucleotides**) in the double helix is specific:
 - A always pairs up with T
 - C always pairs with G**EASY TO COPY!**
- Sequence of nucleotides in the chains varies → **genetic information coding!**

- Genetic information stored in the DNA is copied to an RNA molecule in a process called **transcription**
- RNA directs the protein synthesis by a process called **translation**
- Translation takes place in ribosomes
- The protein's structure, (dictated by the DNA that was copied) determines its function



The Genetic Code

- Every three DNA building blocks (**nucleotides**) correspond to one protein building block: they are called **codons**!
- Codons are faithfully copied into RNA (the "**messenger**") which moves out of the nucleus
- The **codons** copied by the RNA are faithfully translated into proteins by **ribosomes** (in the cytoplasm)
- Each codon corresponds to one protein building block (**amino acid**)



The genetic code: letters that make words!

Codon	Alphabet Letter (amino acid)
GCT	D
GCA	E
TCG	K
ATC	N
AAT	W

DNA building blocks: A, T, C, G

AAT GCA GCA TCG GCA ATC GCT
W E E K E N D word
(protein)

AAT GCA GCT TCG GCA ATC GCT
W E D K E N D nonsense

GCA AAT GCT TCG GCA GCA ATC
E W D K E E N nonsense

It is the order (**sequence**) of codons that produces meaningful words (**functional proteins**)!!

The Genetic Code

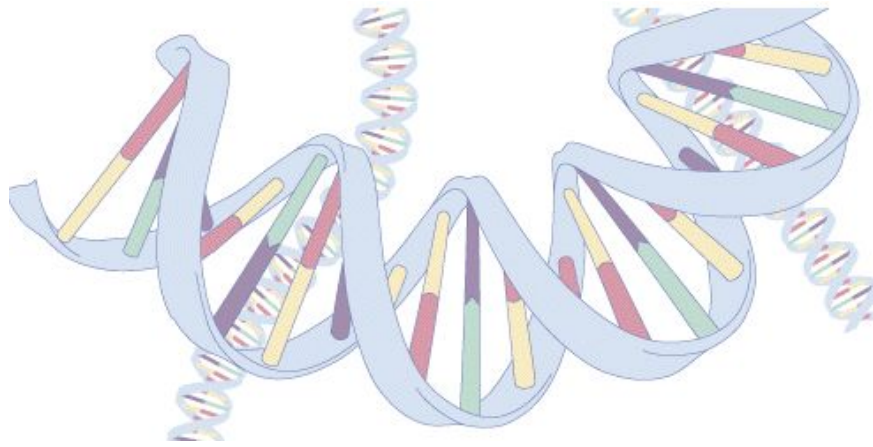
- 26 letters in the alphabet can make for volumes of text!
- 20 amino acids are found in nature
- 4 nucleotides (building blocks) in DNA, hence 64 combinations of unique triplets (**codons**)!
- More than enough to code for all 20 amino acids!!

	1 st	2 nd position				3 rd
	↓	T	C	A	G	↓
T		Phe	Ser	Tyr	Cys	T
		Phe	Ser	Tyr	Cys	C
		Leu	Ser	STOP	STOP	A
		Leu	Ser	STOP	Trp	G
C		Leu	Pro	His	Arg	T
		Leu	Pro	His	Arg	C
		Leu	Pro	Gln	Arg	A
		Leu	Pro	Gln	Arg	G
A		Ile	Thr	Asn	Ser	T
		Ile	Thr	Asn	Ser	C
		Ile	Thr	Lys	Arg	A
		Met	Thr	Lys	Arg	G
G		Val	Ala	Asp	Gly	T
		Val	Ala	Asp	Gly	C
		Val	Ala	Glu	Gly	A
		Val	Ala	Glu	Gly	G

The Genetic Code

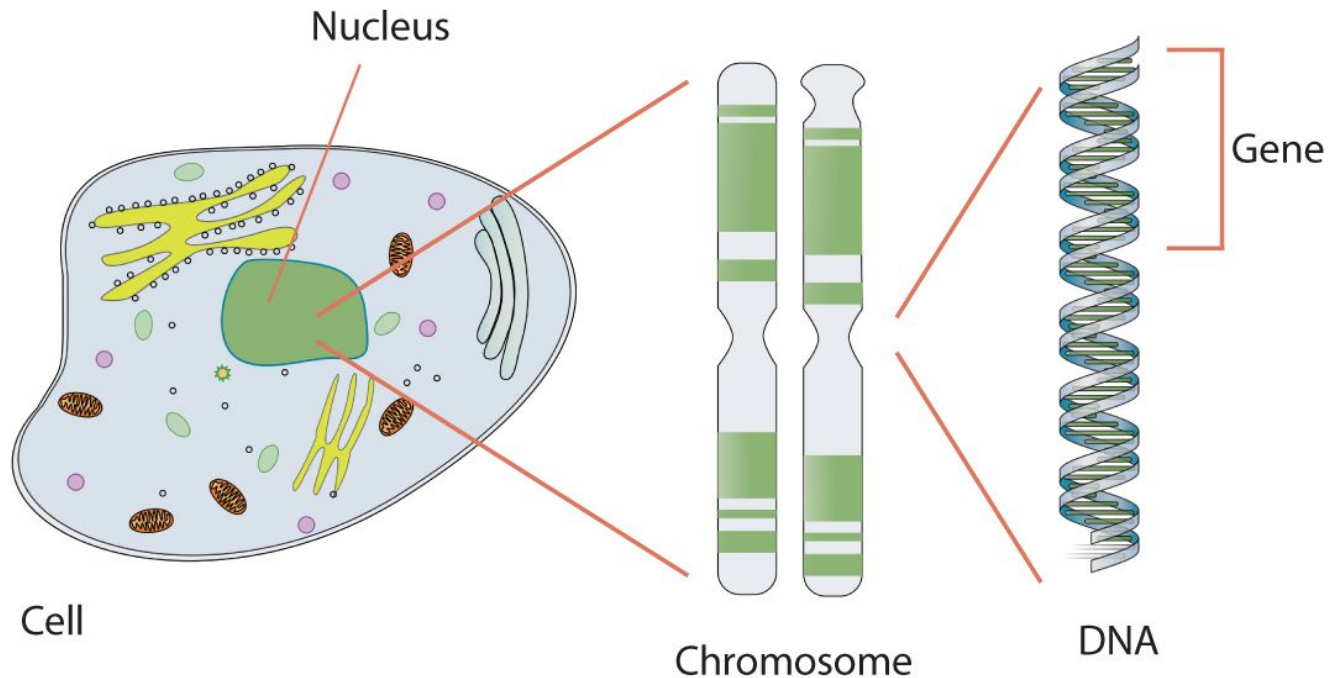
Amino acids

ala - alanine
 arg - arginine
 asn - asparagine
 asp - aspartic acid
 cys - cysteine
 gln - glutamine
 glu - glutamic acid
 gly - glycine
 his - histidine
 ile - isoleucine
 leu - leucine
 lys - lysine
 met - methionine
 phe - phenylalanine
 pro - proline
 ser - serine
 tthr - threonine
 trp - tryptophan
 tyr - tyrosine
 val - valine



Genes, Chromosomes and Genetics

DNA and chromosomes

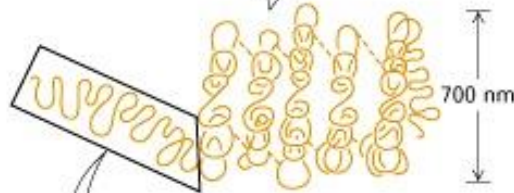


Metaphase chromosome



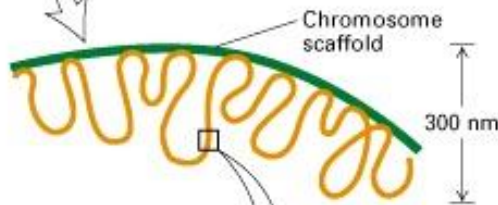
Chromosome

Condensed scaffold-associated form



Condensed scaffold

Extended scaffold-associated form



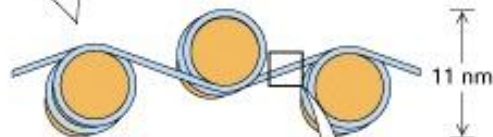
fibers connected to chromosome scaffold

30-nm chromatin fiber of packed nucleosomes



chromatin fibers

"Beads-on-a-string" form of chromatin



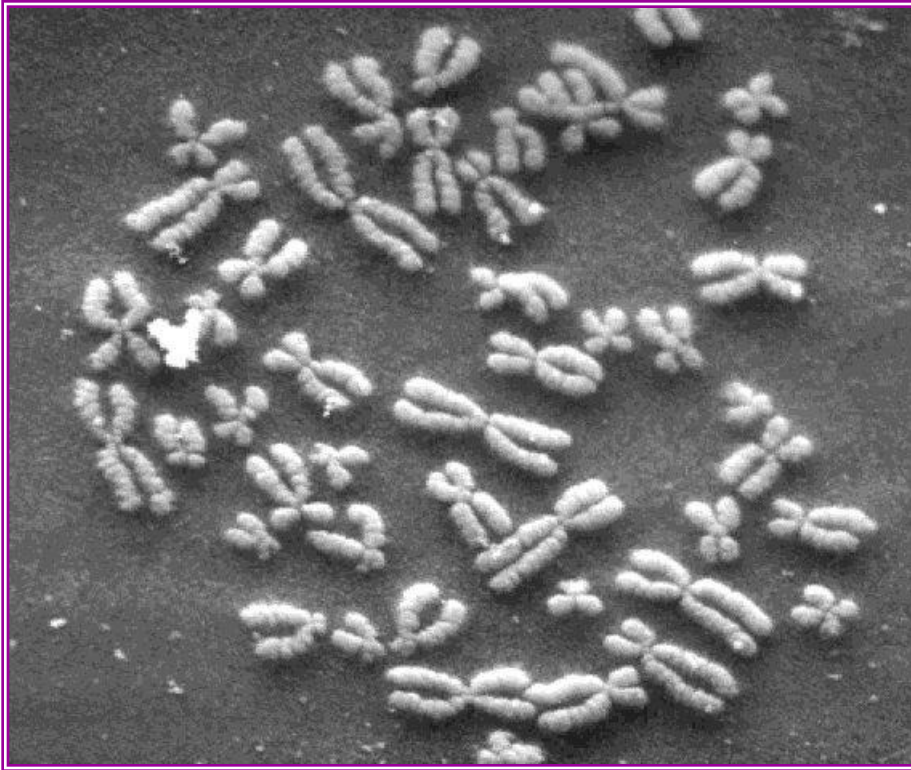
chromatin

Short region of DNA double-helix



DNA

Chromosomes



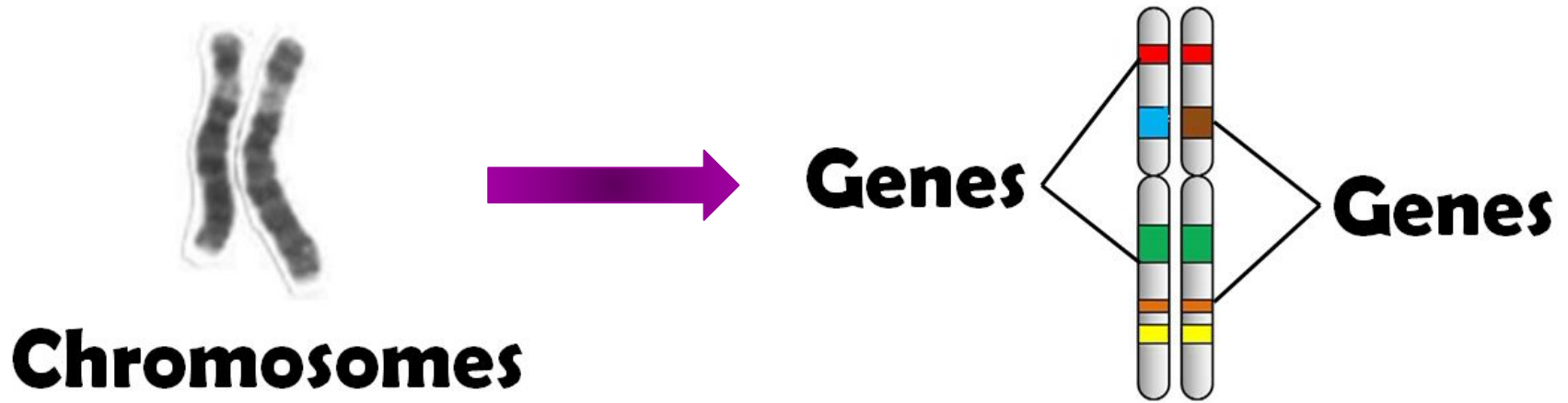
Humans have 23 pairs
of chromosomes, i.e.
**total of 46
chromosomes**

23 chromosomes
from mom

23 chromosomes
from dad

- 2 sex chromosomes:
X and Y
-chromosomes
- XX □ female
- XY □ Male

Chromosomes and genes



A gene: a segment of DNA that codes for the amino acids that make up a specific protein

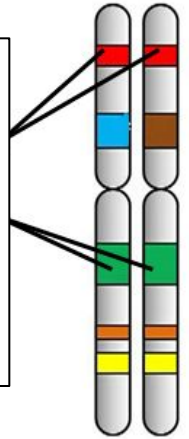
Chromosomes and genes



Chromosomes



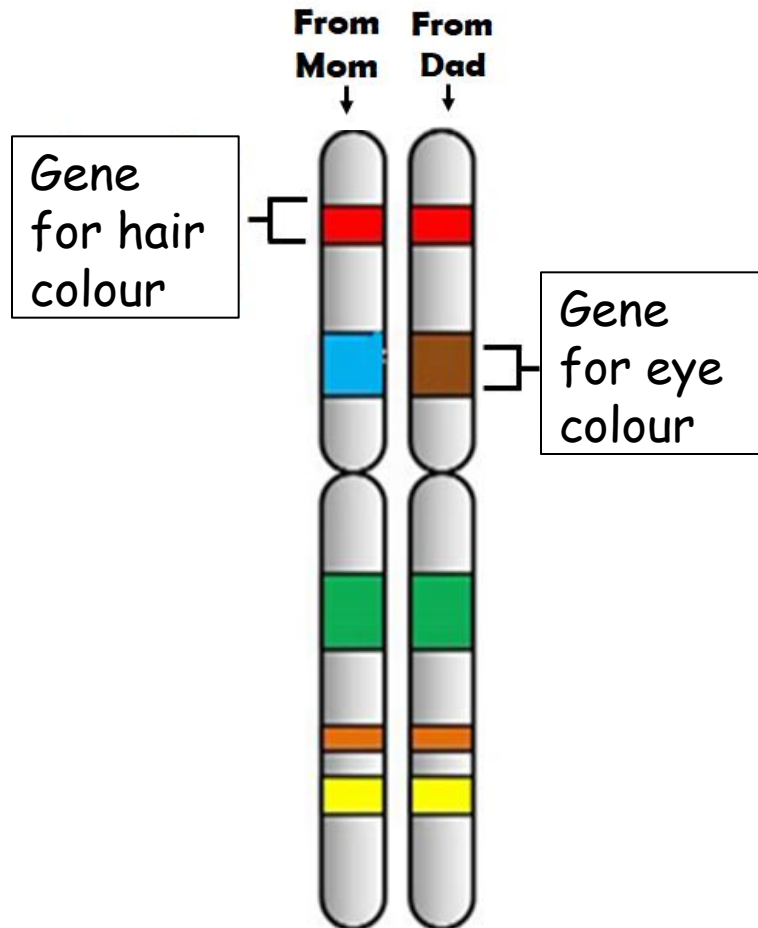
**Each set of genes
codes for a different
protein**



A gene: a piece of DNA that codes for the amino acids that make up a specific protein

Each chromosome contains many genes...

Inheritance of traits



One copy of every gene inherited from Mom

Second copy every gene inherited from Dad

Each copy is called an **allele**

Specific inherited characteristics of an individual (e.g. eye colour, hair colour etc.) that are coded by individual genes are called **traits**.

Traits are *genetically* determined by genes inherited from each parent

Two important terms...

Phenotype: An observable trait in an individual, e.g. hair colour

Genotype: The genetic information that corresponds to this observable trait

Phenotypes

Genotype

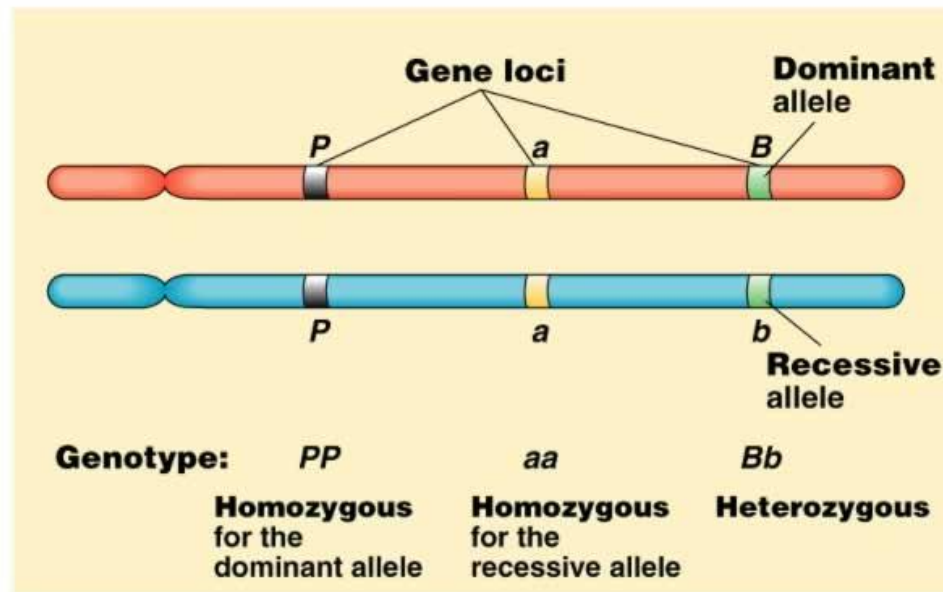
GCCAAGAATGGCTCCACCT
GGCTCTCAGACATTCCCCTGG
TCCAACCCCCAGGCCATCAAG
ATGTCTCAGAGAGGCGGCTAG
ACACCCAGAGACCTCAAGTGA
CCATGTGGGAACGGGATGTTT
CCAGTGACAGGCA



Genotype

ATGTTTCCACCTTCAGGTTCC
ACTGGGCTGATTCCCCCTCC
CACTTTCAAGCTCGGCCCTT
TCAACTCAGAGAGGCGGCTA
GACACCCAGAGACCTCAAGT
GACCATGTGGGAACGGGATG
TTTCCAGTGACAGGCAG

- **Dominant** character: only one allele needed to cause the phenotype (heterozygous)
- **Recessive** character: both alleles needed to cause the phenotype (homozygous)



Some facts about DNA and genes

- About 22 000 genes in man
- Only 2 % of the total DNA in the nucleus (**the genome**) encode proteins (i.e. they are **genes**)
- Genes vary a lot in size:
Humans: average 3000 nucleotides
largest 2.4 million nucleotides
- 50 % of DNA in chromosomes is repetitive DNA ("junk DNA")

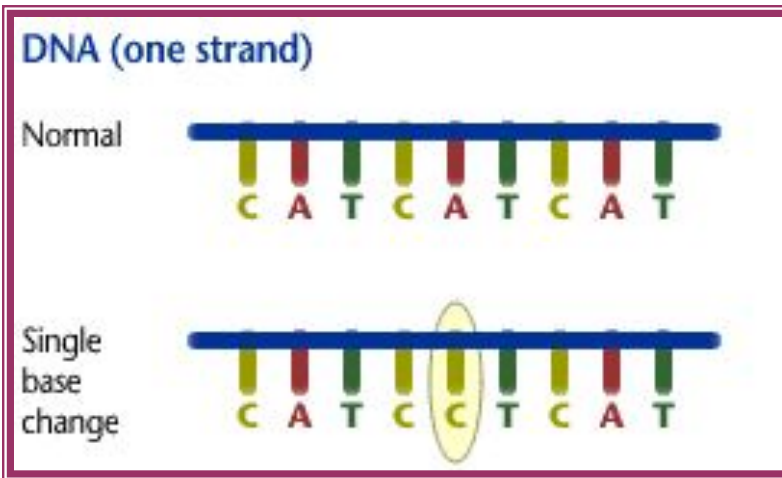
Mutations

- Alterations in DNA sequence
- Caused by chemical and physiological agents and errors in **DNA replication** (i.e. when cell divide)
- Cells can repair some mistakes
- If not repaired, changes in DNA sequence are made permanent by **DNA replication**
- A mutation can have no effect (majority of cases), or it can have a **deleterious** effect (e.g. damage a gene)

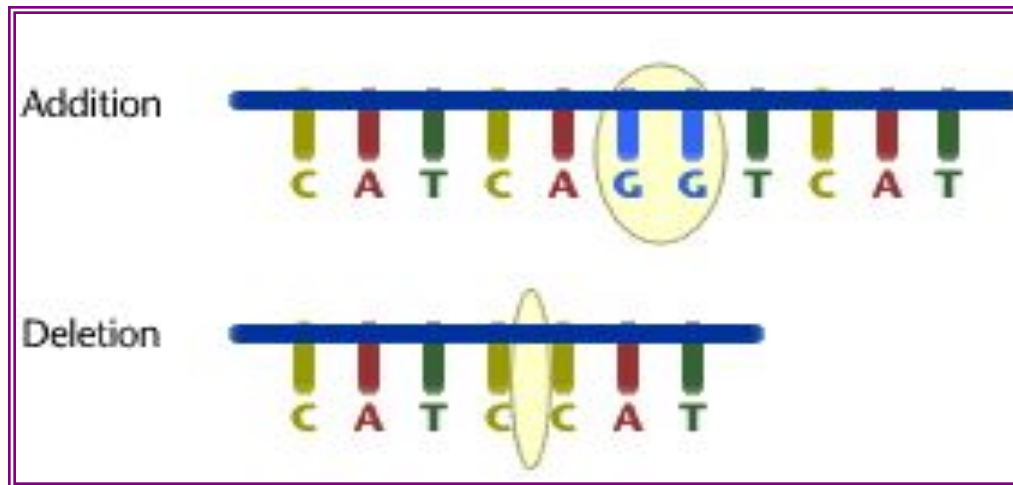


Point mutations:

Single nucleotide mutations:



1. **Missense mutation:** leads to an amino acid change
2. **Silent mutation:** does not change the amino acid
3. **Nonsense mutation:** causes premature stop-codon



- **Frameshift mutations:**
 - insertion/deletion
 - dublication
 - translocation
- Change the oder of the codons, hence the identity of the translated protein
- Severe impacts on protein structure

Inherited diseases

- DNA mutations are significant in development of diseases
- Inherited diseases are caused by mutations passed from a parent to a offspring
- **Monogenic diseases:** disease is caused by one mutation in one gene
- **Multifactorial diseases:** disease is caused by interaction of different mutations and environmental factors



Questions? Contact me: john.strouboulis@kcl.ac.uk

Basic Genetics

[View Teach.Genetics for Classroom Materials](#)

MESSAGE FROM LEARN GENETICS

LEARN GENETICS VISITORS

We're asking for your help. For over 20 years, the Learn Genetics website has provided engaging, multimedia educational materials at [no cost](#).

Learn Genetics is one of the most-used science websites. Tens of millions of visitors come to our site each year to find the science and health information they're looking for.

If Learn.Genetics is useful to you, **please** take a moment to **donate** - even a few dollars from each of our visitors would add up to a significant amount.

Your support will help us keep Learn Genetics free and available to everyone. It will also help us develop new content for you.

Please help us keep Learn.Genetics going!

Thank you,

The Genetic Science Learning Center team - creators of Learn.Genetics

MORE ABOUT CHROMOSOMES



HOW DO SCIENTISTS READ CHROMOSOMES?

To "read" a set of chromosomes, scientists look for key features to identify their similarities and differences.

[Learn more](#)

TOUR OF BASIC GENETICS



WHAT ARE TRAITS?

Explore traits, the characteristics that make us unique.

[video](#)



WHAT ARE DNA & GENES?

Get to know the molecule that holds the instructions for building every living thing.

[video](#)



WHAT ARE PROTEINS?

Learn about proteins, the molecules we're made of.

[video](#)



WHAT ARE CHROMOSOMES?

Learn about the relationships between chromosomes, genes, and DNA molecules.

[video](#)



WHAT IS INHERITANCE?

Learn how traits pass from parents to offspring.

[video](#)



WHAT IS MUTATION?

Take a look at how variation occurs.

[video](#)

MORE ABOUT TRAITS

Excellent website for understanding basic genetics:
<https://learn.genetics.utah.edu/content/basics/>