22-Genetics

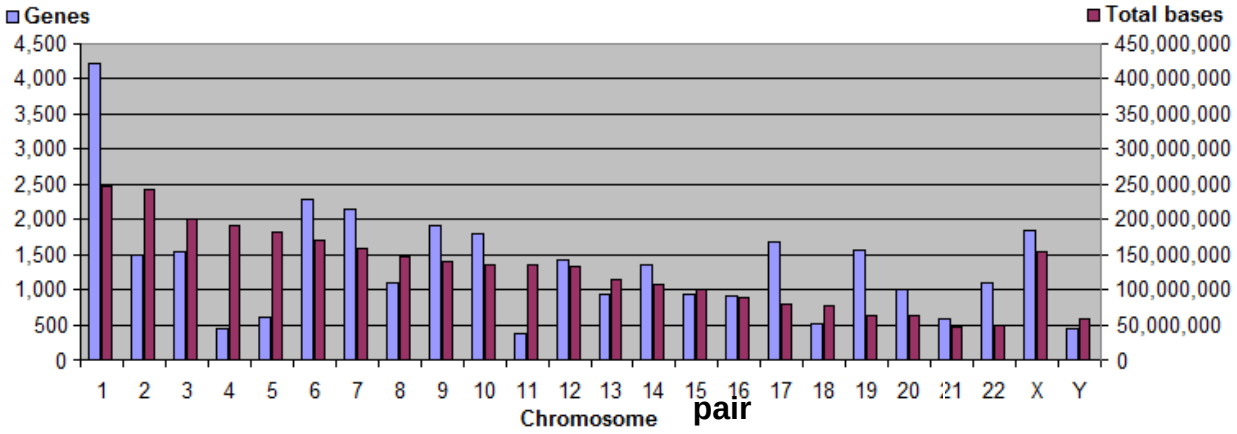
A- Intro What you need to know about chromosomes:

1) A **chromosome** is an organized structure of DNA (hereditary molecule) and protein that is found in cells.

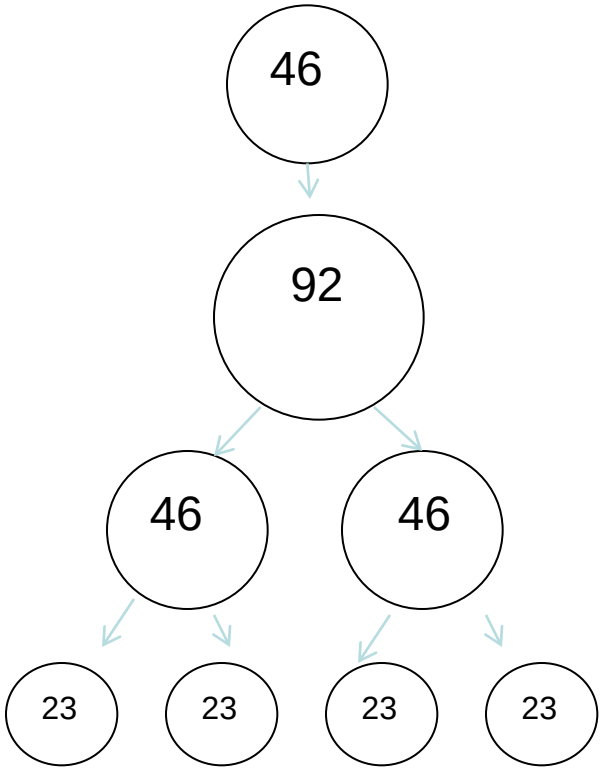
2) In humans, chromosomes come in 23 pairs. So there are 46 chromosomes in all.

3) Pairs of chromosomes have similar but not necessarily identical genes. A gene has information to make one type of protein.

4) Each sex cells forms from one member of each chromosome pair, so a sex cell only has 23 chromosomes in all.



Meiosis



1. Factors Responsible for Biological Character Traits

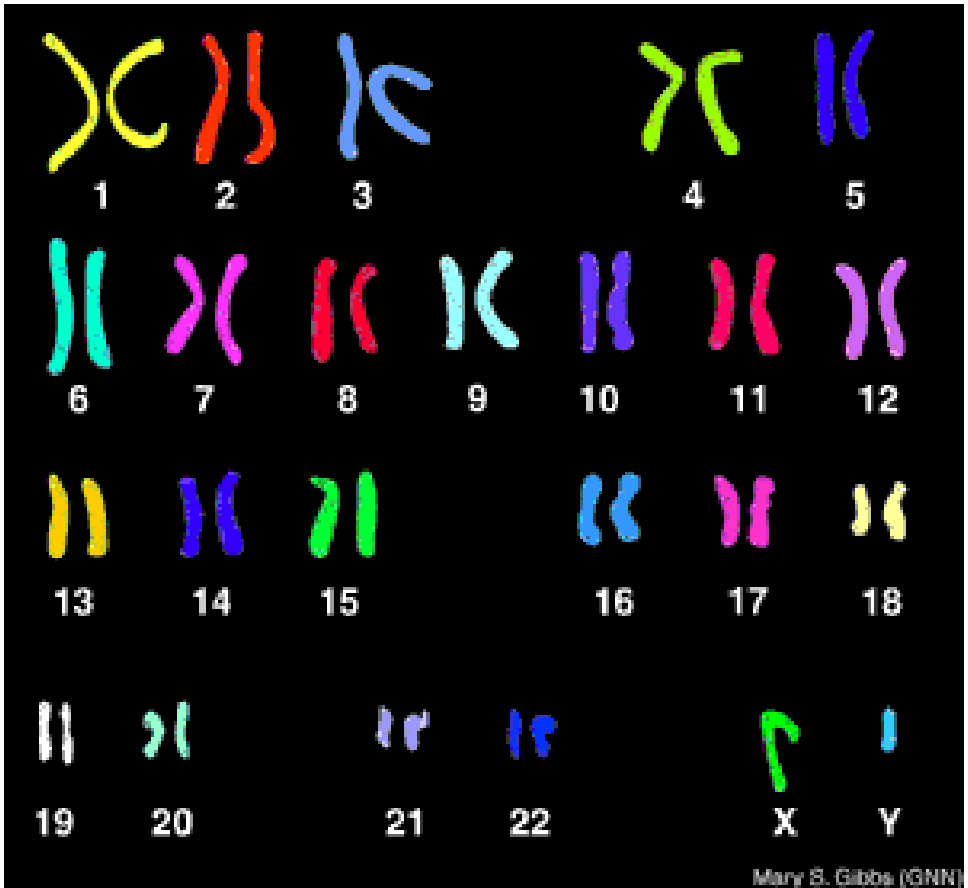
What are character traits?

single features or quantifiable measurements of an organism

What is responsible for those traits?

Genetic information

The structure of chromosomes during mitosis



1.1 Distinguish between:

a) Chromatin material:

Mass of genetic material composed of DNA and proteins that condense to form chromosomes during eukaryotic cell division

b) Chromosome:

a threadlike strand of DNA in the cell nucleus that carries the genes in a linear order. Has proteins too.

c) Chromosome pair

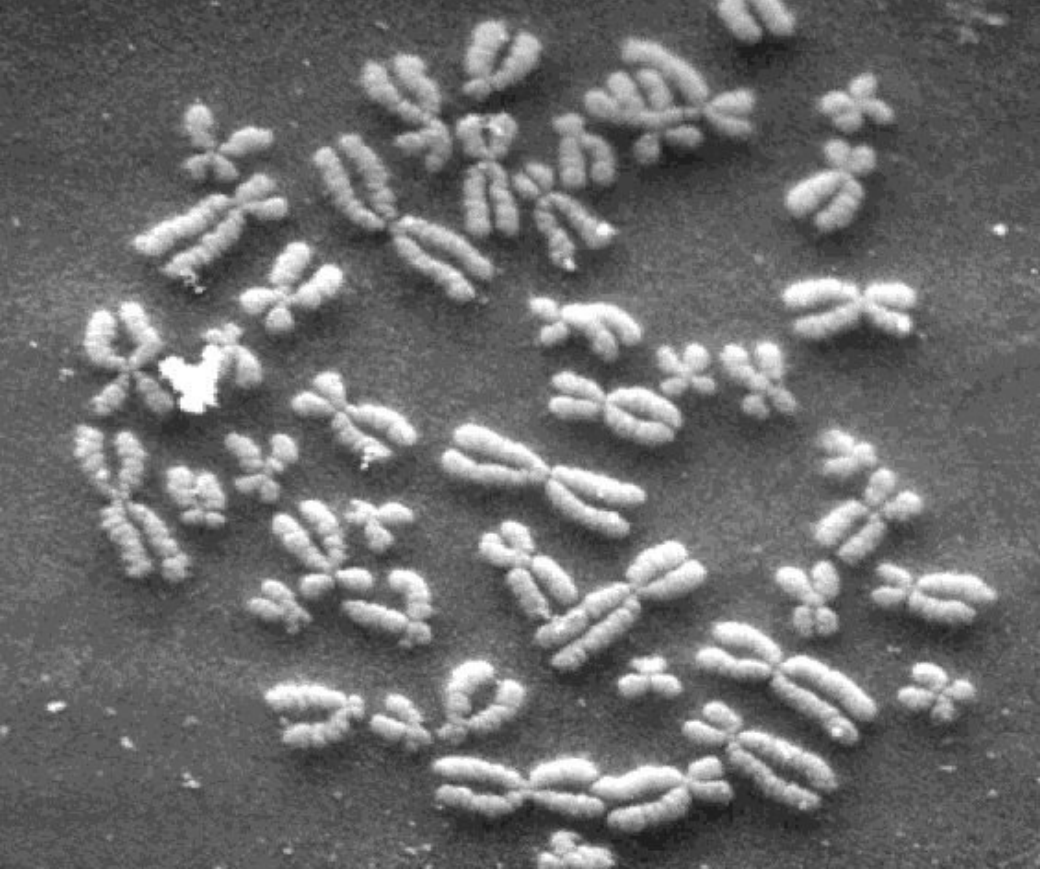
Two chromosomes. 23 such pairs in humans

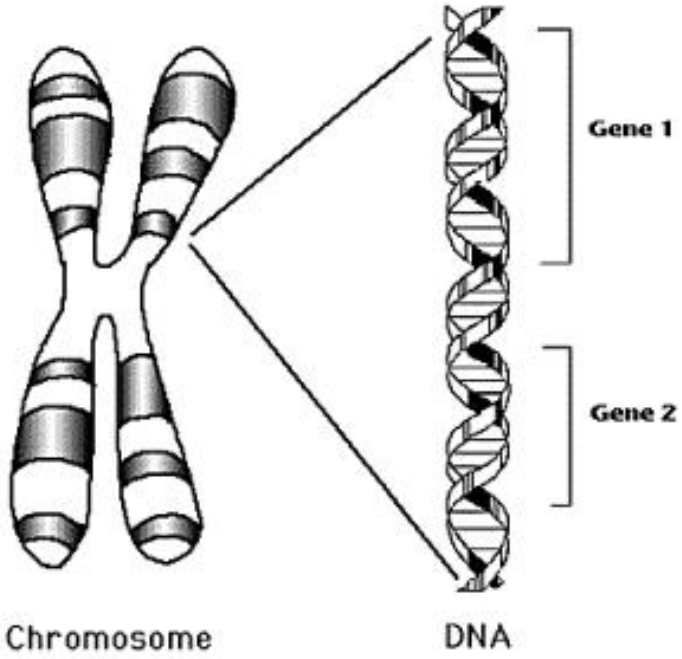
d) DNA

deoxyribonucleic acid

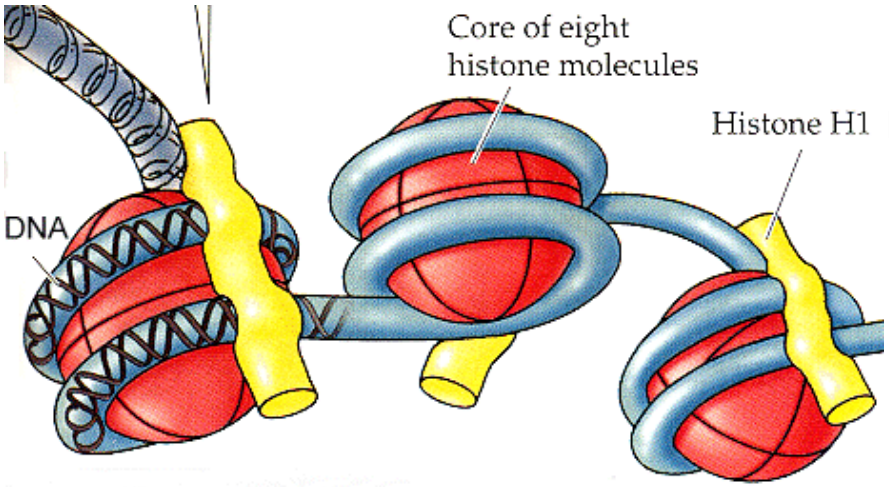
e) Gene:

a section of DNA that codes for 1 protein. Sometimes leads to a trait.



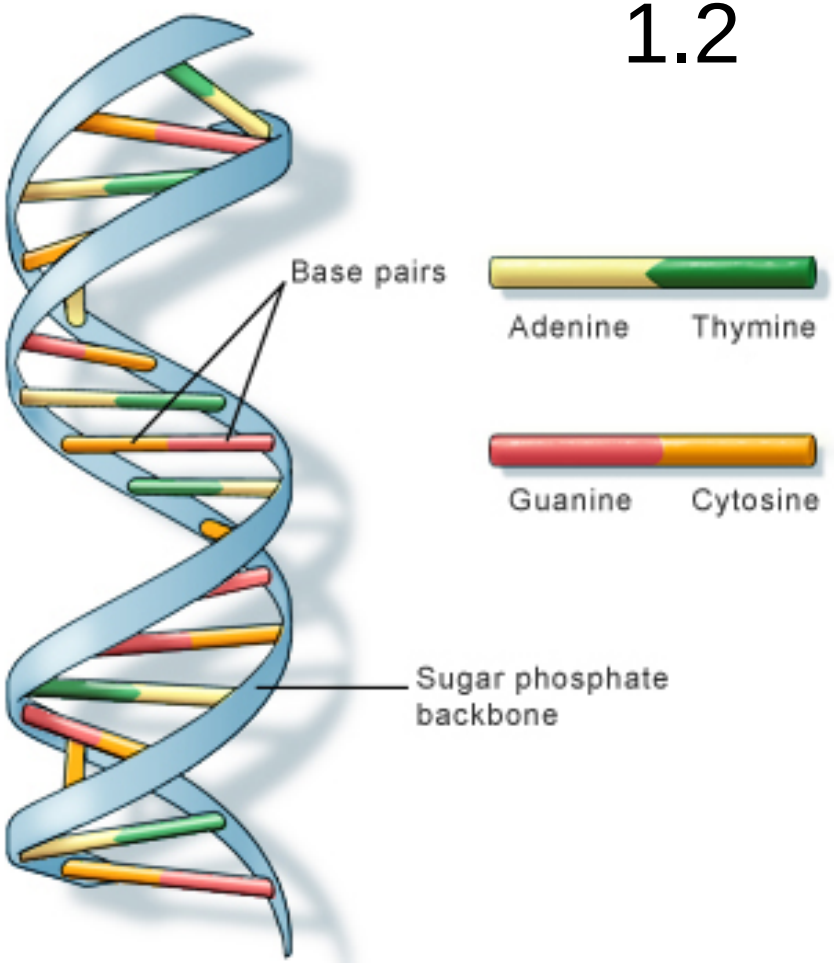


Genes

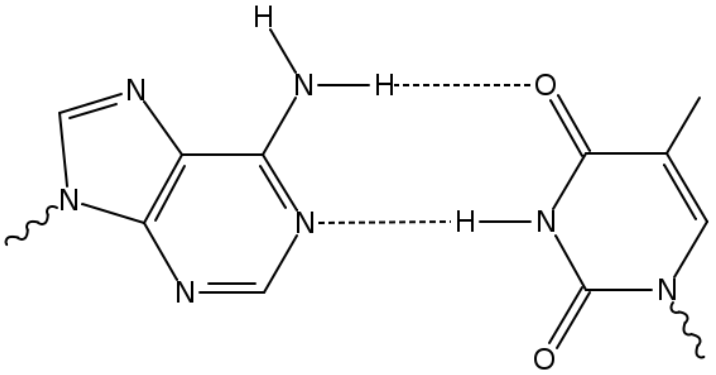


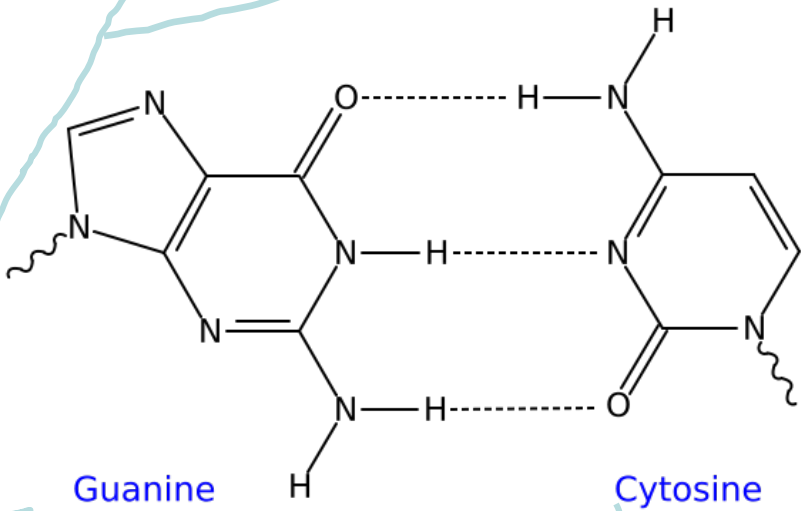
How many human genes are there? 20 000 -25 000
Original estimate: 100 000

1.2 DNA



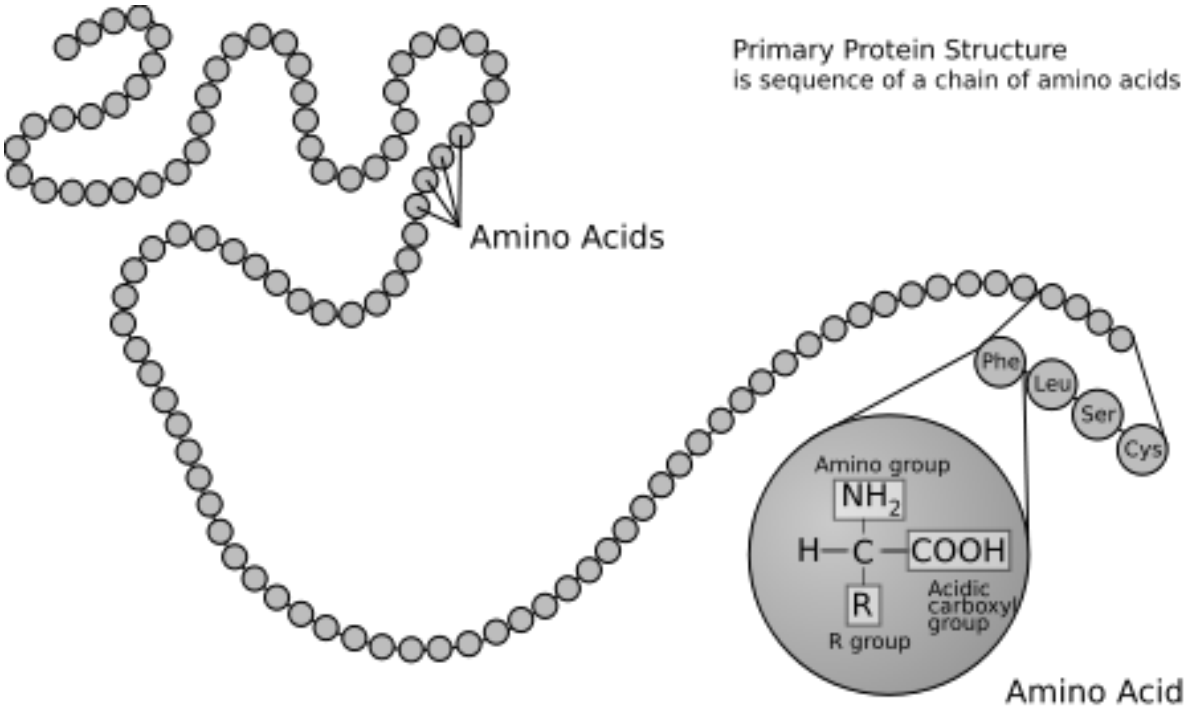
U.S. National Library of Medicine





What is DNA? A molecule consisting of nucleotide bases, sugar and phosphates

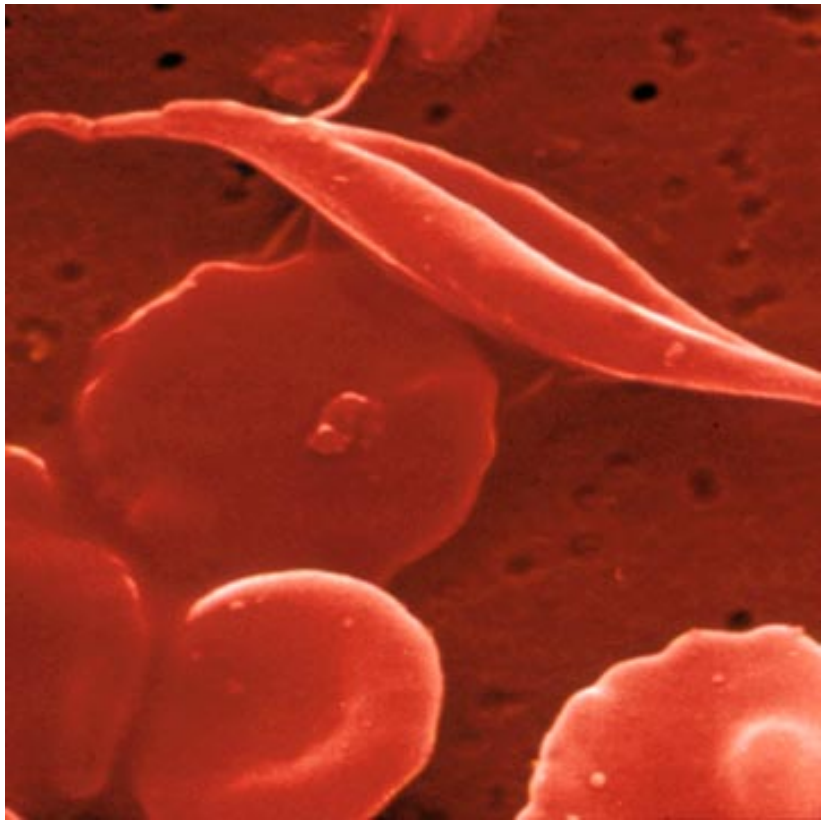
1.3a Protein



What is a protein?

A group of amino acids usually with a 3d shape that plays an important role in its function.

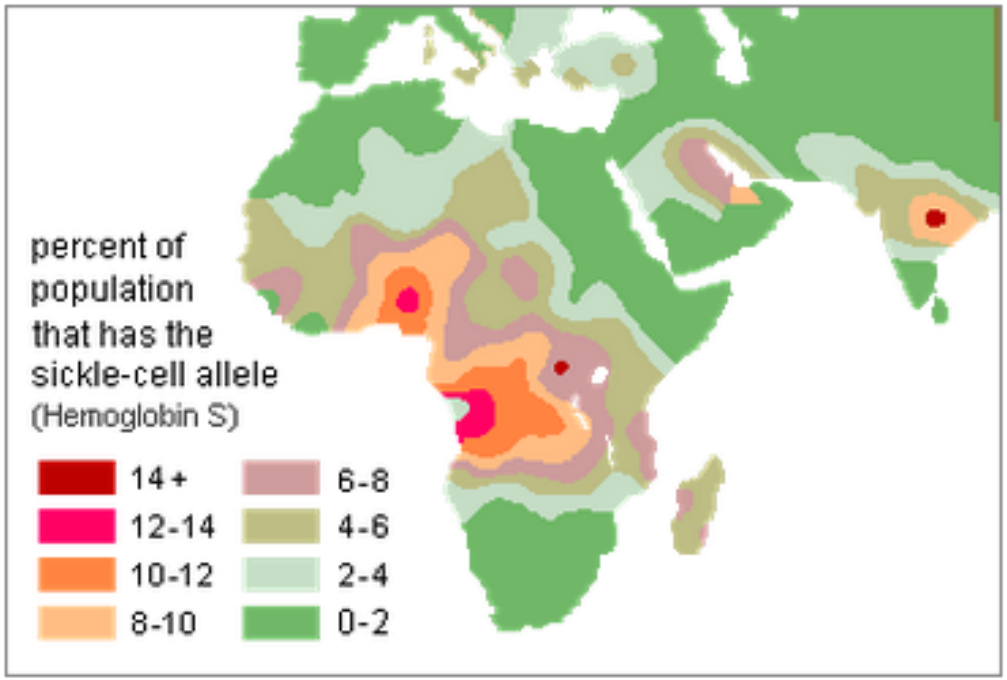
Proteins can act as enzymes (molecules that speed up reactions), structural materials, chemical messengers(hormones) and antibodies.

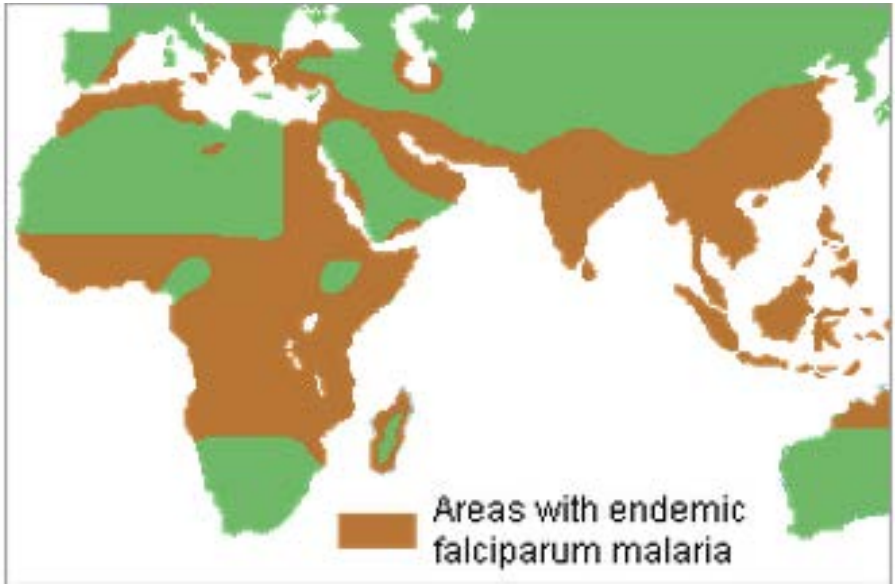


Normal blood cells and sickled cells.
The latter have hemoglobin (a protein) with one wrong amino acid in the sequence. What went wrong?

A change in the genetic code(mutation)
occurred and it was distributed in parts of the human population.

Why is there a higher percent of the sickle cell allelic gene in certain areas?





1.3b RNA

What is RNA? How does it differ from DNA?

RNA = ribonucleic acid
-does not form double helix
-contains different sugar
-has uracil instead of thymine pairing with adenine(important)
-usually relays genetic information instead of storing it(even more important)
-some forms can act as enzymes

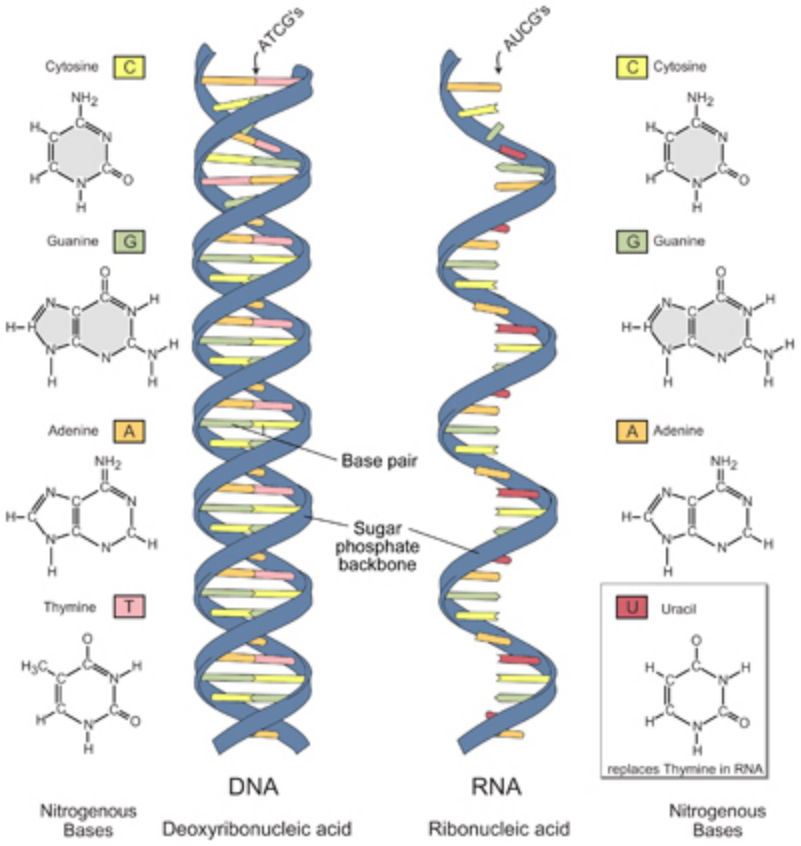
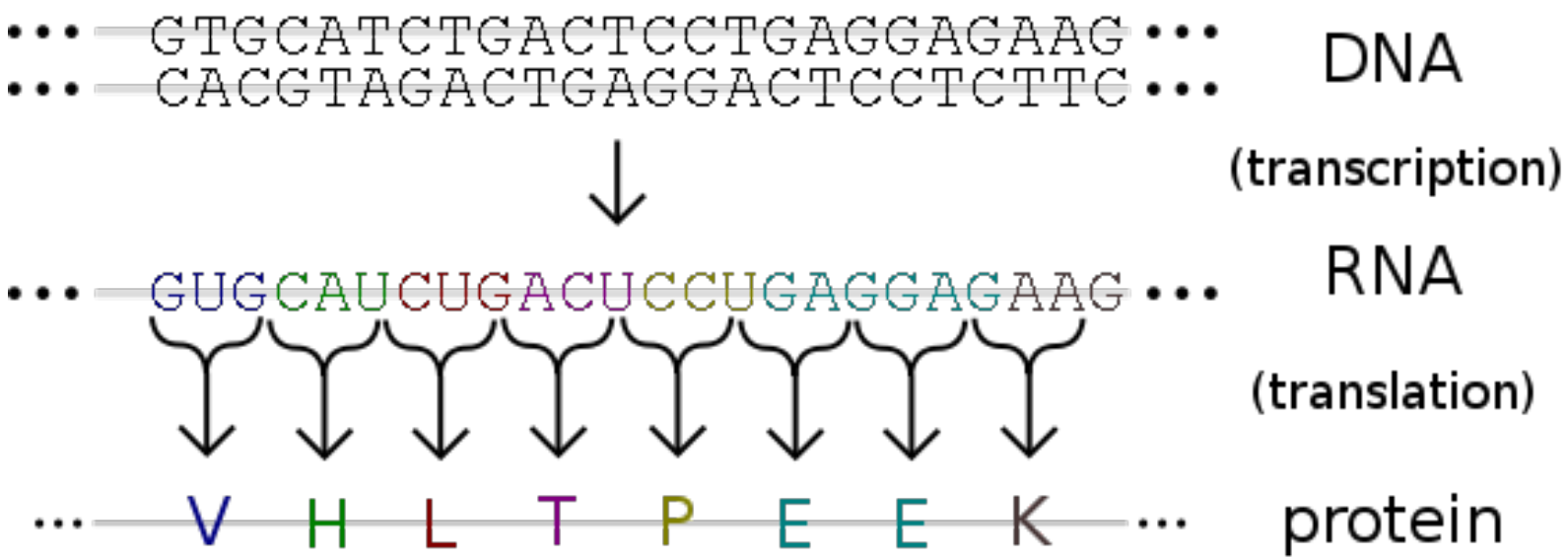
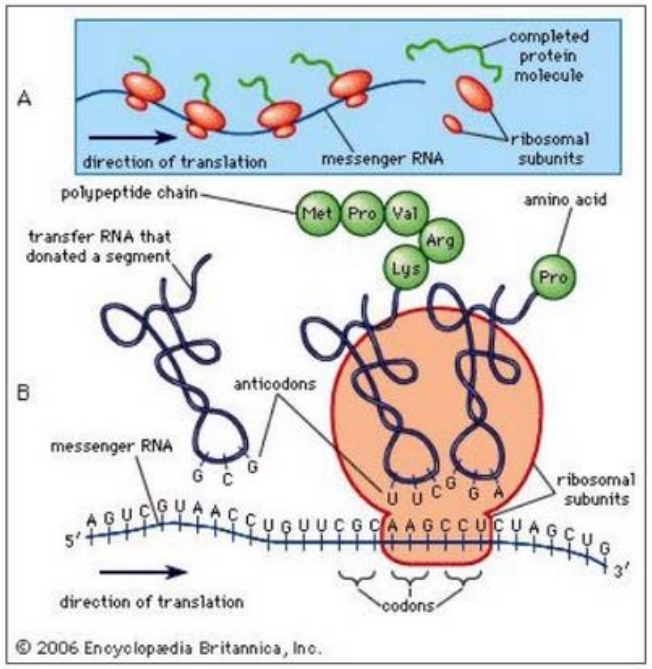


Image adapted from: National Human Genome Research Institute.

1.4 Protein Synthesis





Use the architect-foreman analogy to explain protein synthesis.

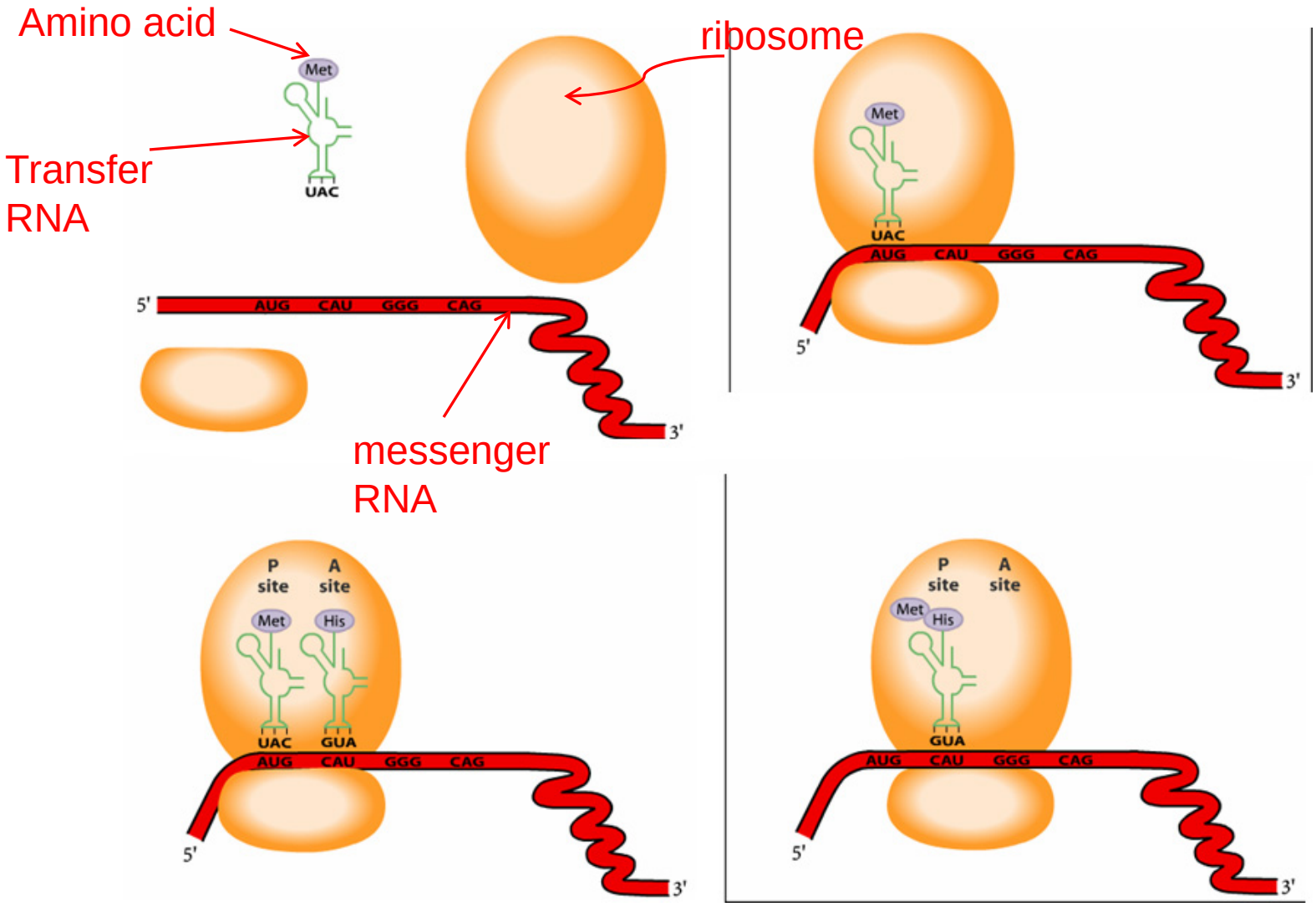
DNA is like the architect; it has the plan for building
mRNA is the foreman who goes to work site with the plan
tRNAs are the workers who assemble the building according to the plan.

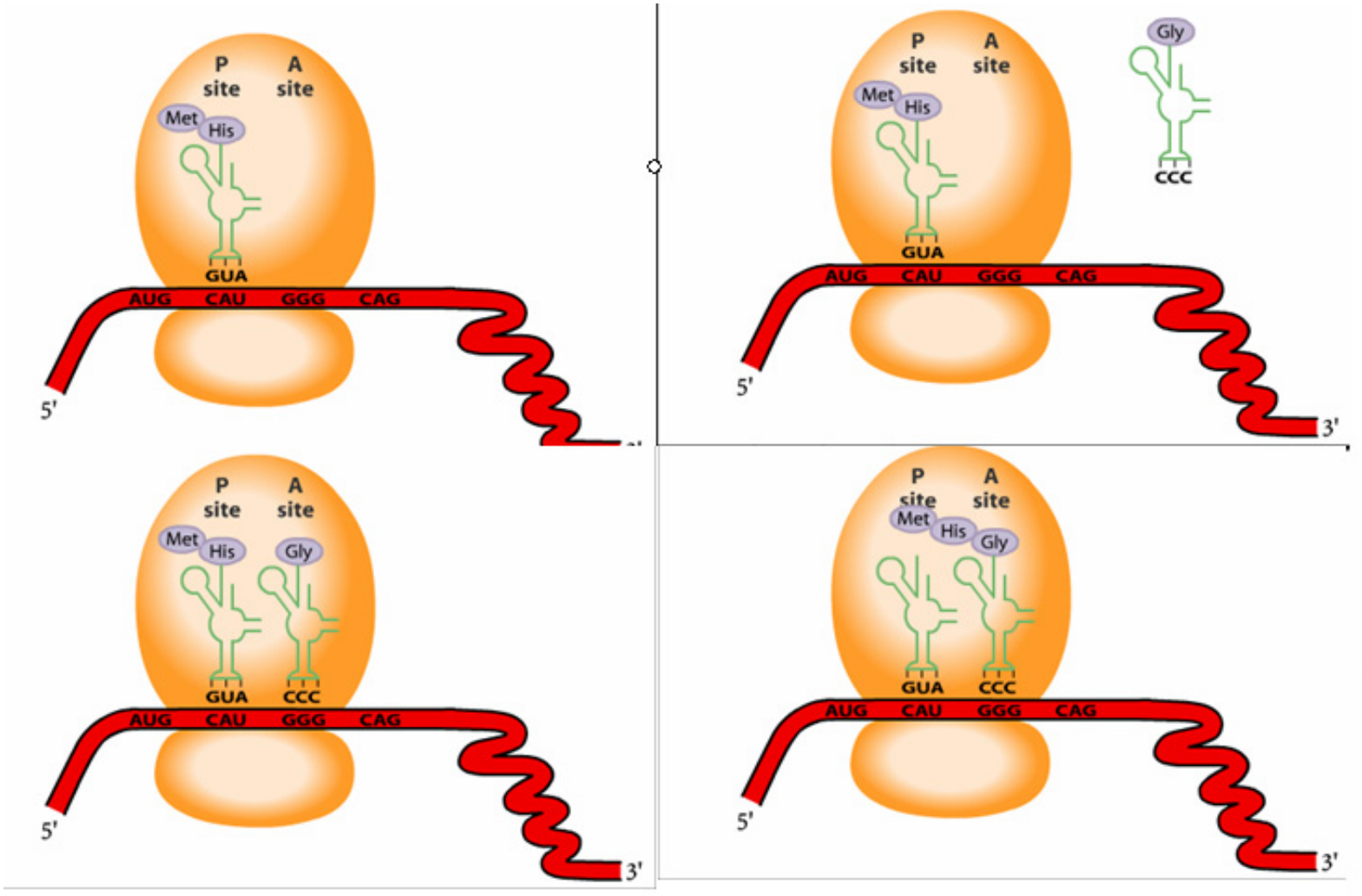
b) Label each structure or molecule and describe its role

A gene(part of DNA)is a series of codes to make a protein.
Each three bases(letters) code for 1 of the amino acids.
mRNA forms on the surface of DNA(essentially copying the code) and then moves to the ribosome.
tRNA collects amino acids and they get connected according to the sequence on mRNA.

Protein Synthesis

Label each structure or molecule and describe its role




P site
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AUG CAU GGG CAG
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3'
Gly
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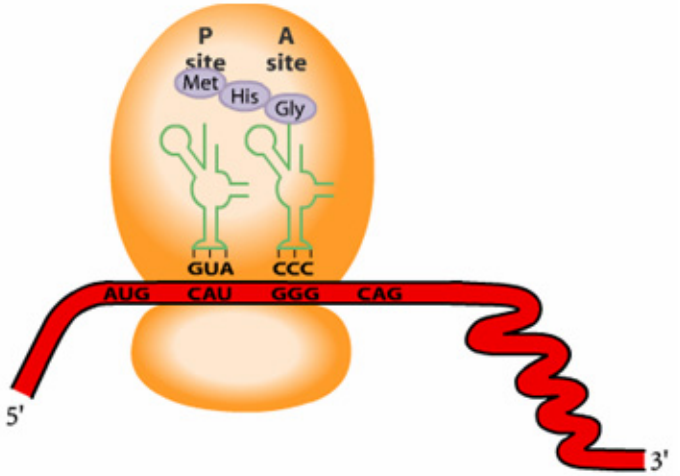
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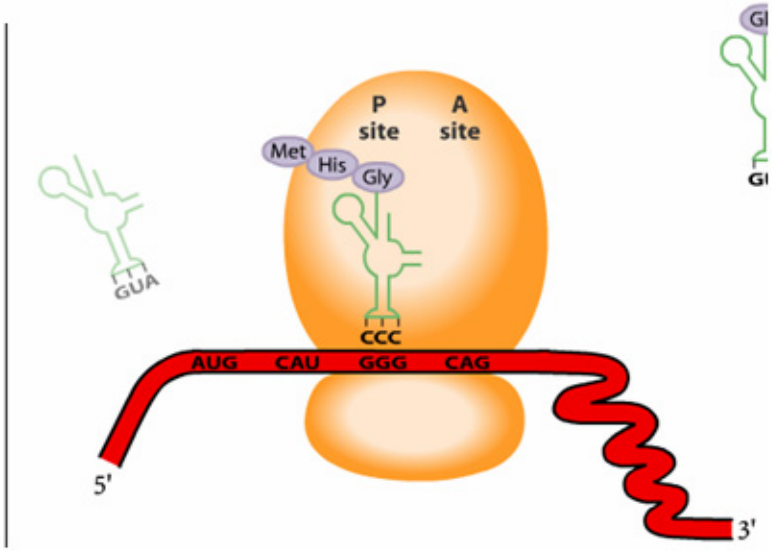
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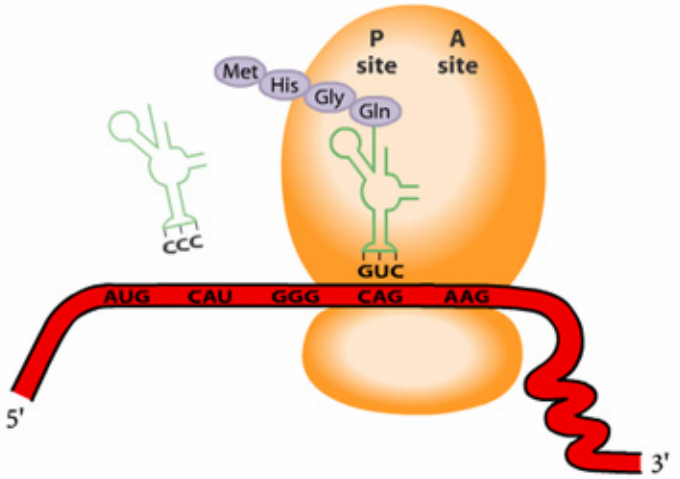
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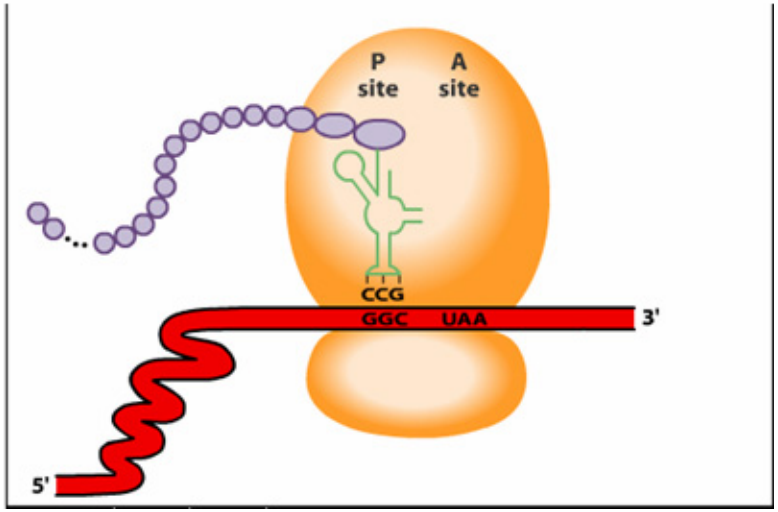
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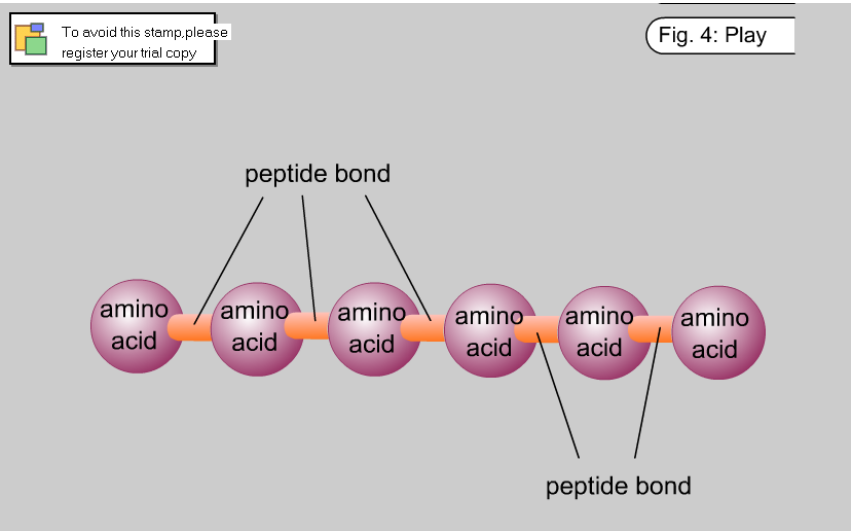


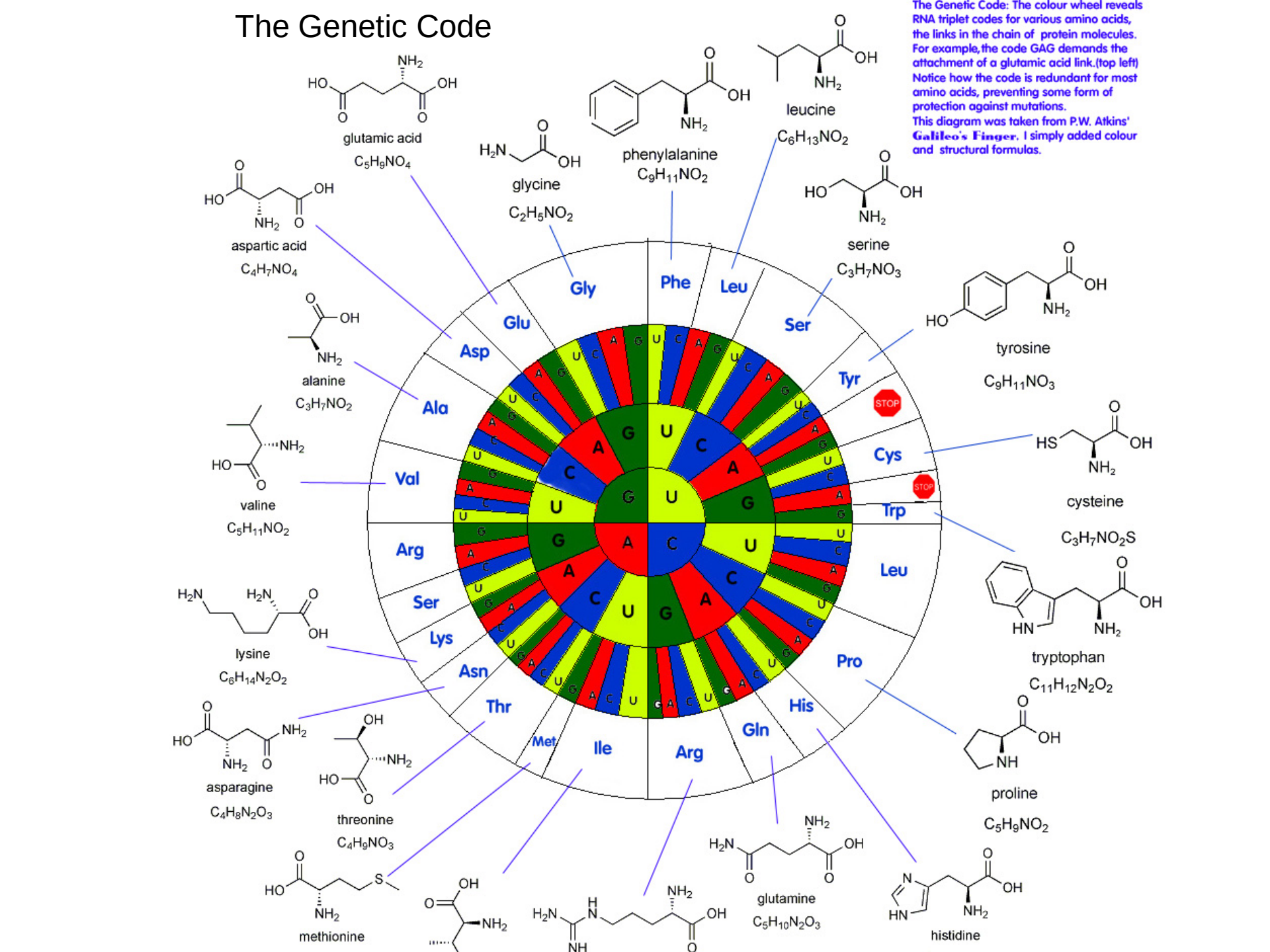
Summary: the gene has the information needed to make 1 protein . The info is in the form of DNA base triplets. Through, A-U and G-C matching, a complementary version of this info is transferred to messenger RNA molecules. Messenger RNA moves to the ribosome where transfer RNA molecules pick up specific amino acid molecules and link them in the order dictated by messenger RNA.

Analogy: DNA is like the architect, messenger RNA the foreman, transfer RNA is the Worker and protein is the product. Different proteins have different sequences of amino acids.

The triplet sequence of the transfer RNA's strongly resembles that of DNA except that In DNA there is thymine(T) instead of uracil(U).

The Genetic Code

The Genetic Code: The colour wheel reveals RNA triplet codes for various amino acids, the links in the chain of protein molecules. For example, the code GAG demands the attachment of a glutamic acid link.(top left) Notice how the code is redundant for most amino acids, preventing some form of protection against mutations.
This diagram was taken from P.W. Atkins' **Galileo's Finger**. I simply added colour and structural formulas.



p156 Use the messenger-RNA codes on p158 to obtain the amino acids that would be assembled if the m-RNA code was the following:

UUUACUCGC

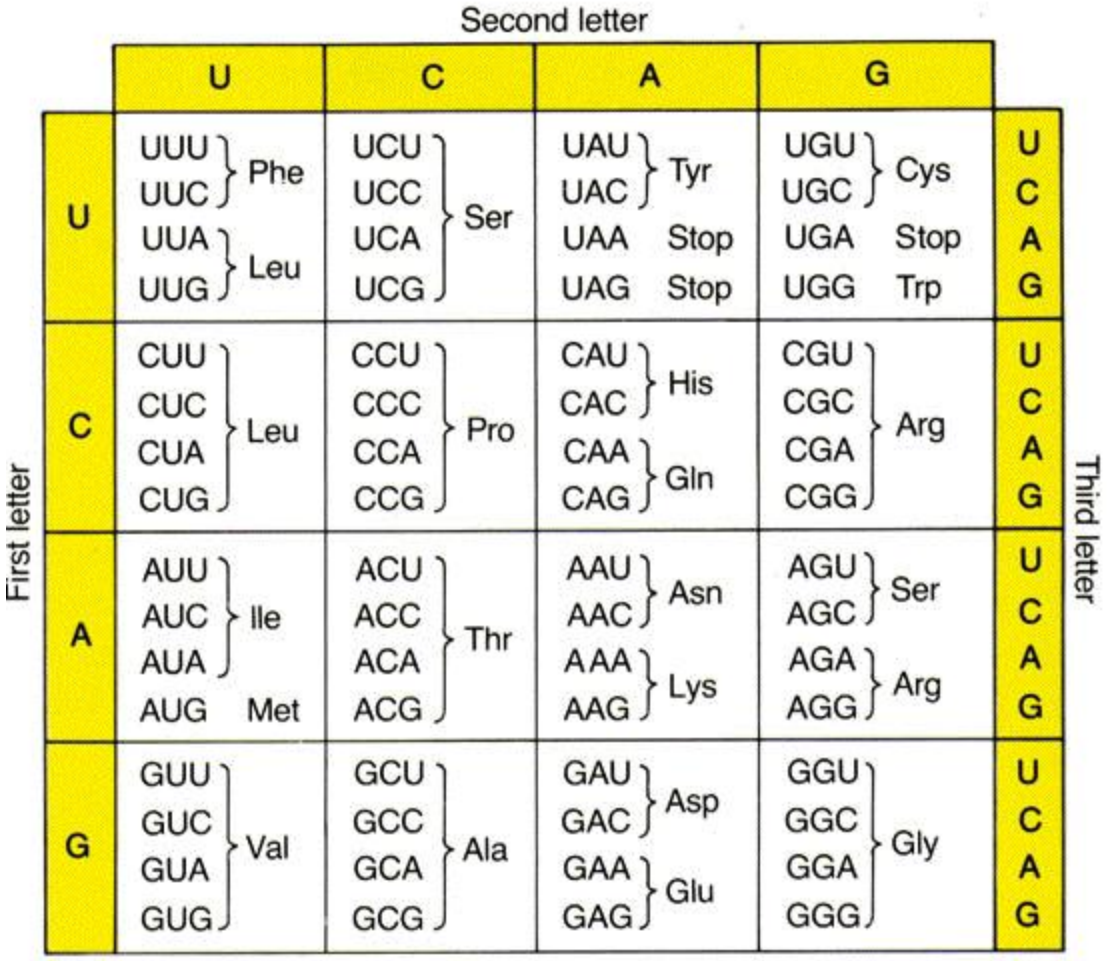
Phe-Thr-Arg

What would be the DNA codes corresponding to the above messenger RNA codes?

AAATGAGCG

Transfer RNA codes?

AAA and UGA and GCG



Why is there often more than 1 code for the same amino acid?

It's protection against specific types of mutations, which are changes in the genetic code.

Diseases Caused By Mutations

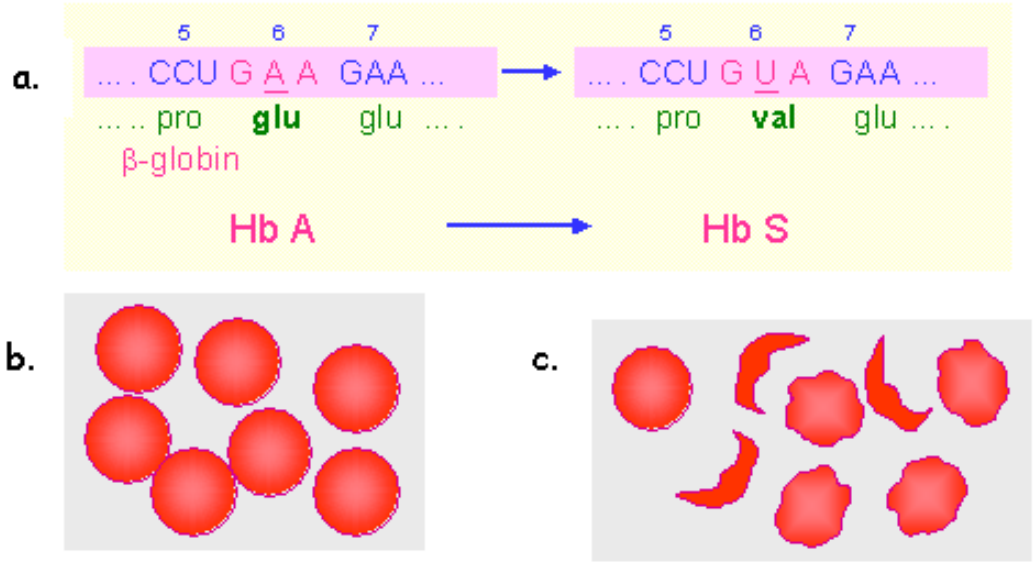


Fig. 5.1 (a) Point mutation in codon number six of the beta β-globin gene results in the substitution of the amino acid number glutamine with valine and the formation of haemoglobin S (HbS); (b) Red blood cells in a smear of normal blood containing HbA; (c) crenated and sickle-shaped red blood cells in sickle cell anaemia.

More Diseases Caused By Mutations

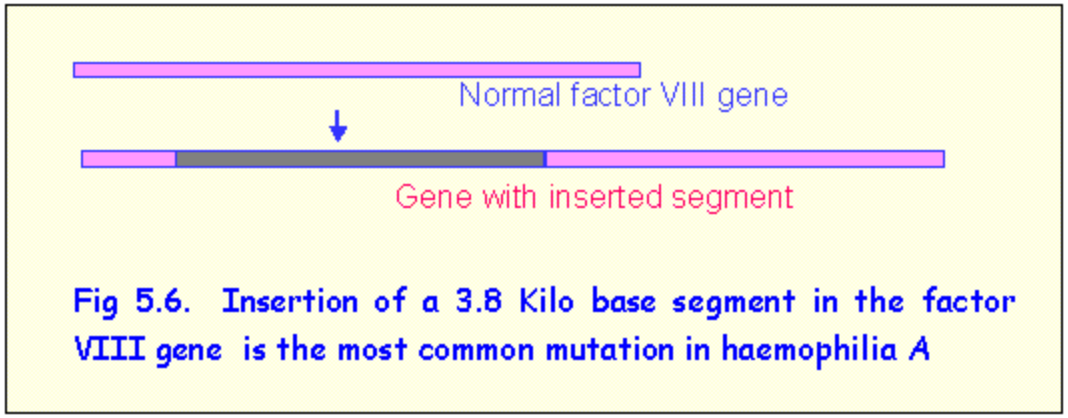


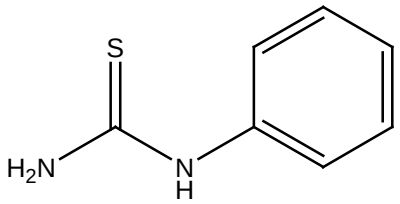
Fig 5.6. Insertion of a 3.8 Kilo base segment in the factor VIII gene is the most common mutation in haemophilia A

Cancer usually results from a series of mutations within a single cell. Often, a faulty, damaged, or missing p53 gene is to blame. The p53 gene makes a protein that stops mutated cells from dividing.

Examples of Good Mutations

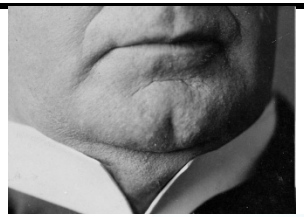
1. a specific 32 base pair deletion in human CCR5 (CCR5-Δ32) gives HIV resistance to homozygotes (2 copies of mutation) and delays AIDS onset in heterozygotes (have only one copy of mutation)
2. The same mutation may have protected people who had the altered gene during the Bubonic Plague.

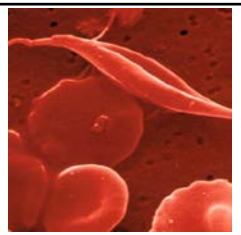
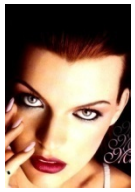
2. Principles of Heredity
A. Mendelian Traits

Ability to taste phenylthiocarbamide(PTC, a bitter compound)	 phenylthiocarbamide
Albinism (recessive)	
Blood type	A,B,AB,O
Brachydactyly (Shortness of fingers and toes)	

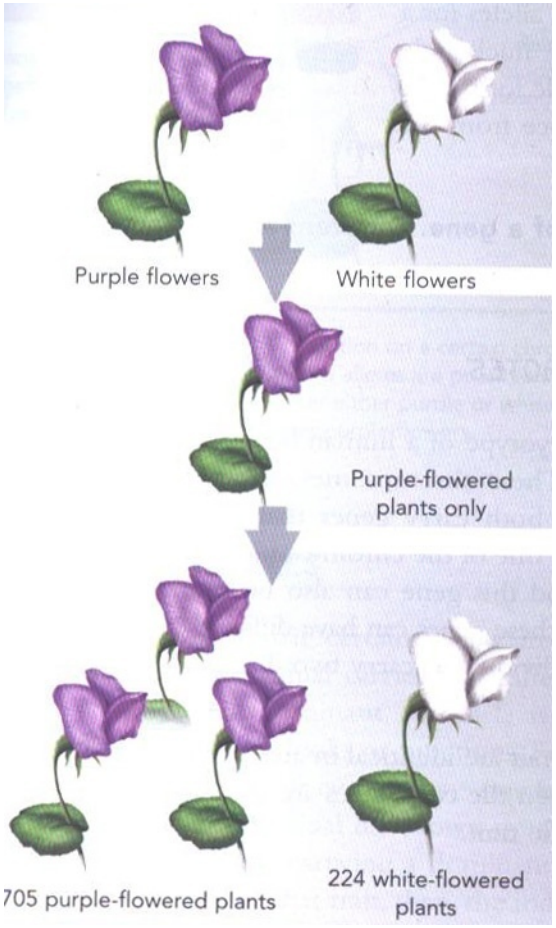
When is a trait considered "Mendelian"?

More Mendelian Traits

Cleft chin (dominant)	 
Cheek dimples (dominant)	
Free (dominant) or attached (recessive) earlobes	
Wet (dominant) or dry (recessive) earwax	

Face freckles (dominant)	
Hitchhiker's thumb (recessive)	
Sexdactyly (Six fingers/toes)	
Sickle-cell trait (also considered co-dominant)	
Widow's peak (dominant)	 

Examples of Crossbreeding
(Crossbreeding is the exchange of gametes (sex cells and their DNA) between two different individuals during sexual reproduction)



Example 1

How do you explain the results of Mendel's experiments with pea plants?

So- called Mendelian characteristics are controlled by two genes, one from each of a pair of chromosomes. For a dominant trait to be expressed, only one gene has to be present. If a trait is recessive, then two copies of the gene must exist.
Here, the purple allelic gene P
(allele= one of two from a pair)
Is dominant over the white allelic gene p.

Dominant allele	Recessive allele	Phenotypes	Genotypes
P	p	Purple flowers 	PP or Pp
		White flowers 	pp
Y	y	Yellow seeds 	YY or Yy
		Green seeds 	yy
R	r	Round shape 	RR or Rr
		Wrinkled shape 	rr
L	l	Long stem (about 3 m) 	LL or Ll
		Short stem (about 30 cm) 	ll

Example 2

If a pea plant that is heterozygous for stem length is crossed with a short-stemmed one, what % of the offspring will be short when fully grown?

Ll X ll

♂

	L	l
l	Ll	ll
l	Ll	ll

Two *ll* out of 4 possibilities:

50% will be short

Example 3

a) If you grow pea plants from green seeds for several plant generations, will you ever get yellow-seeded plants, assuming no mutations? No green is recessive(see chart)

b) What is meant by “assuming no mutations”?

A mutation is a change in the genetic code caused by radiation or chemicals(natural or artificial) This in theory could cause a gene for green to mutate into a yellow one.

Blue American lobster (Homarus americanus). Taken at the New England Aquarium (Boston, MA, December 2006. Copyright © 2006

Dihybrid Cross

In summer squash, white fruit color (W) is dominant over yellow fruit color (w) and disk-shaped fruit (D) is dominant over sphere-shaped fruit (d).. If a squash plant true-breeding for white, disk-shaped fruit is crossed with a plant true-breeding for yellow, sphere-shaped fruit, what will the phenotypic and genotypic ratios be for the 1st and 2nd generations?

WWDD X wwdd

	WD	WD	WD	WD
wd	WwDd	WwDd	WwDd	WwDd
wd	WwDd	WwDd	WwDd	WwDd
wd	WwDd	WwDd	WwDd	WwDd
wd	WwDd	WwDd	WwDd	WwDd

F_2 = second generation

WwDd X WwDd

	WD	Wd	wD	wd
WD	WWDD	WWDd	WwDD	WwDd
Wd	WWDd	WWdd	WwDd	Wwdd
wD	WwDD	WwDd	wwDD	wwDd
wd	WwDd	Wwdd	wwDd	wwdd

Phenotypic ratios:

White and disk-shaped 9/16: 56.25%

White and sphere-shaped 3/16: 18.75%

Yellow and disk-shaped 3/16: 18.75%

Yellow and sphere-shaped 1/16: 6.25%

Genotypic ratios:

WwDd **WwDD** **WWDd** **WWDD** **Wwdd** **WWdd** **wwDd** **wwDD** **wwdd**

4	2	2	1	2	1	2	1	1