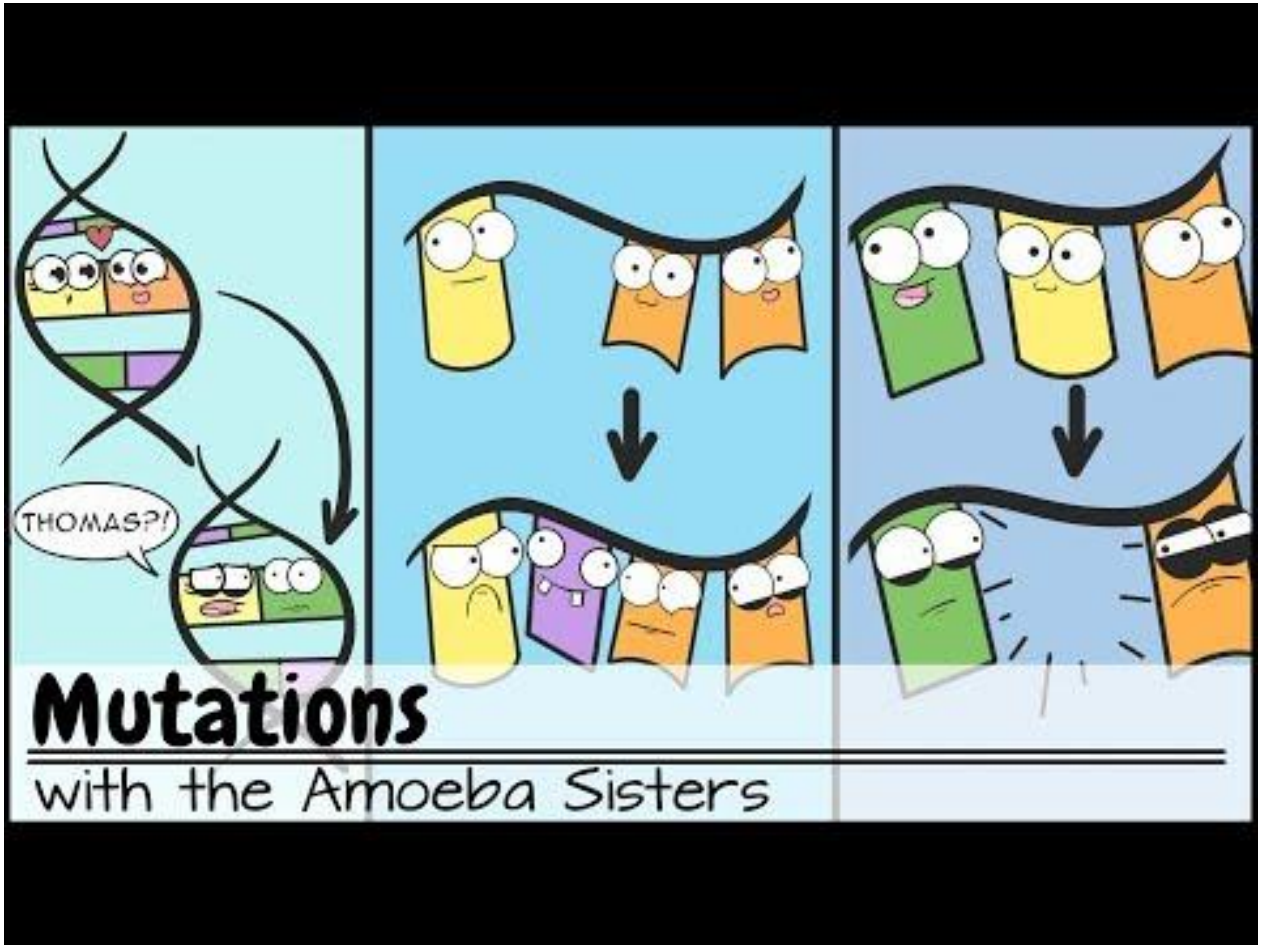
6.2 Mutations

SBI3U


THOMAS?!
Mutations
with the Amoeba Sisters

Mutation: a change in the genetic code

- Can have a positive, neutral or negative effect on phenotype

  Some mutations result in cell death and do not get replicated while others do not result in death and are replicated as cells divide.

Caused by :

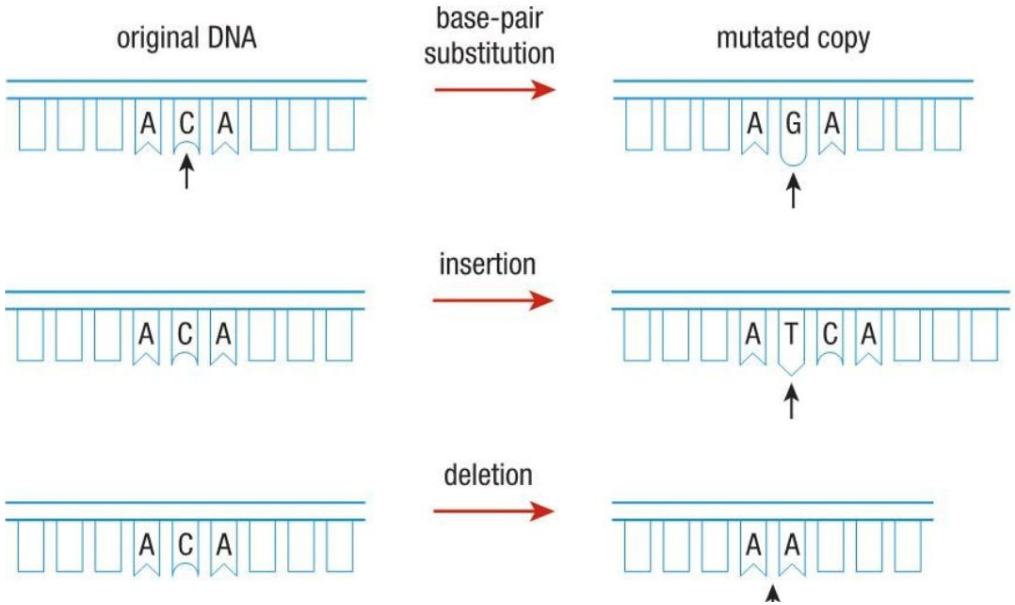
- Environmental agents (radiation, chemicals)
- Errors during cell division

Types of mutations

- Point mutations
- Chromosome mutations

1. Point mutations

Point mutation: a **small-scale** change in the nitrogenous base sequence of DNA

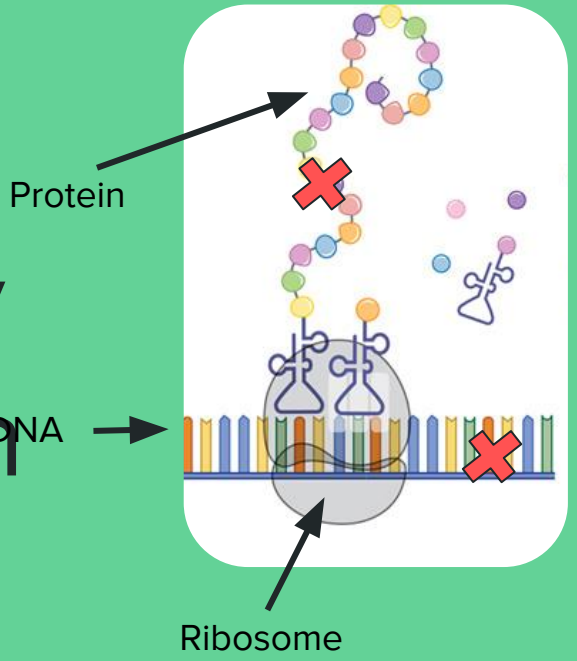


Base-pair substitution - nucleotide is replaced by a different nucleotide

Insertion - nucleotide is added

Deletion - nucleotide is eliminated

Altered DNA sequences are read by ribosomes and result in the production of altered proteins.

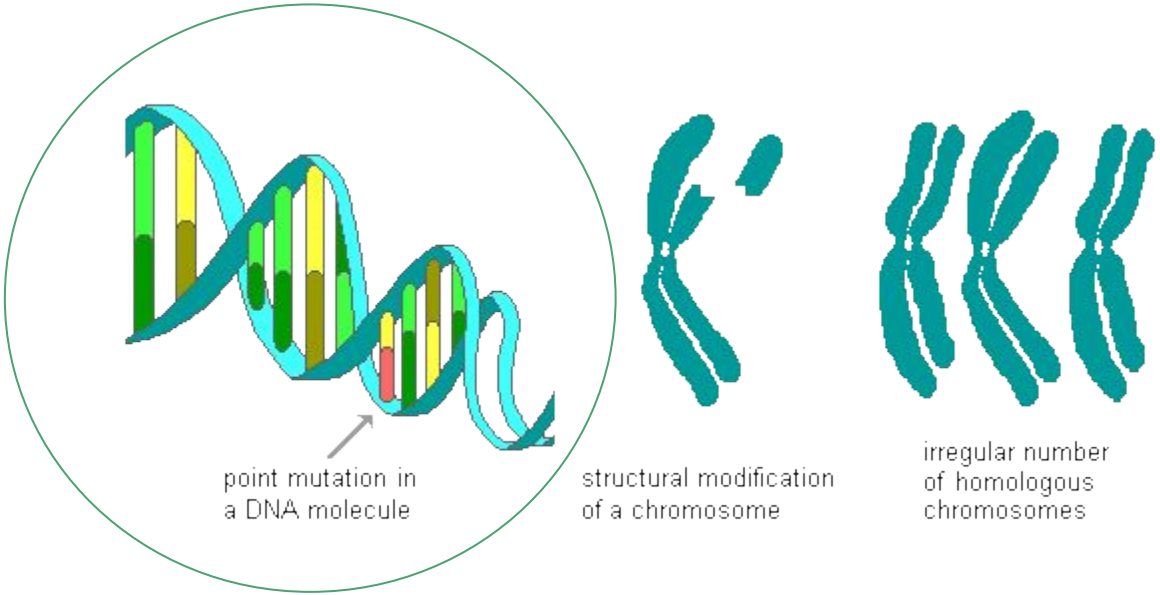


2. Chromosome Mutations

Chromosome mutation: an error that involves an entire chromosome or a large part of a chromosome.

- **Having too few or too many chromosomes due to improper separation of chromosomes or sister chromatids**
  - E.g. Down syndrome, Edwards syndrome, etc.

- **Large scale deletions, insertions or inversions of portions of chromosomes**



Compared to a point mutation, **chromosome mutations** affect a larger portion of the chromosome or chromosome number.

Inheriting Mutations

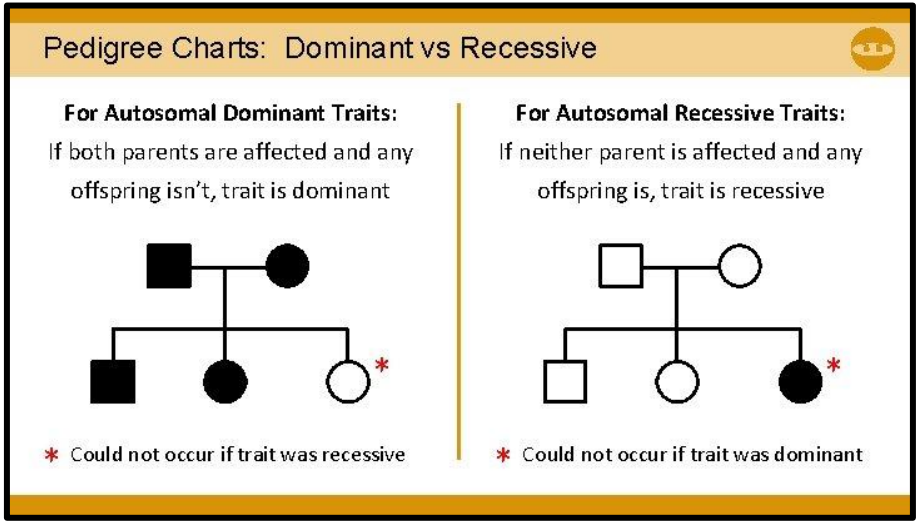
- Mutations that occur in the somatic cells are not passed down to offspring

- Mutations that occur in the sex cells (gametes) are passed down. Offspring will have the mutation in every cell of its body
  - Offspring will be able to pass it down to next generation

Mutations that Occur in Gametes

If the mutation is **autosomal dominant,** it could be expressed in the first generation and every generation after that.

If the mutation is **recessive,** it may not be expressed for many generations and reappear later on.

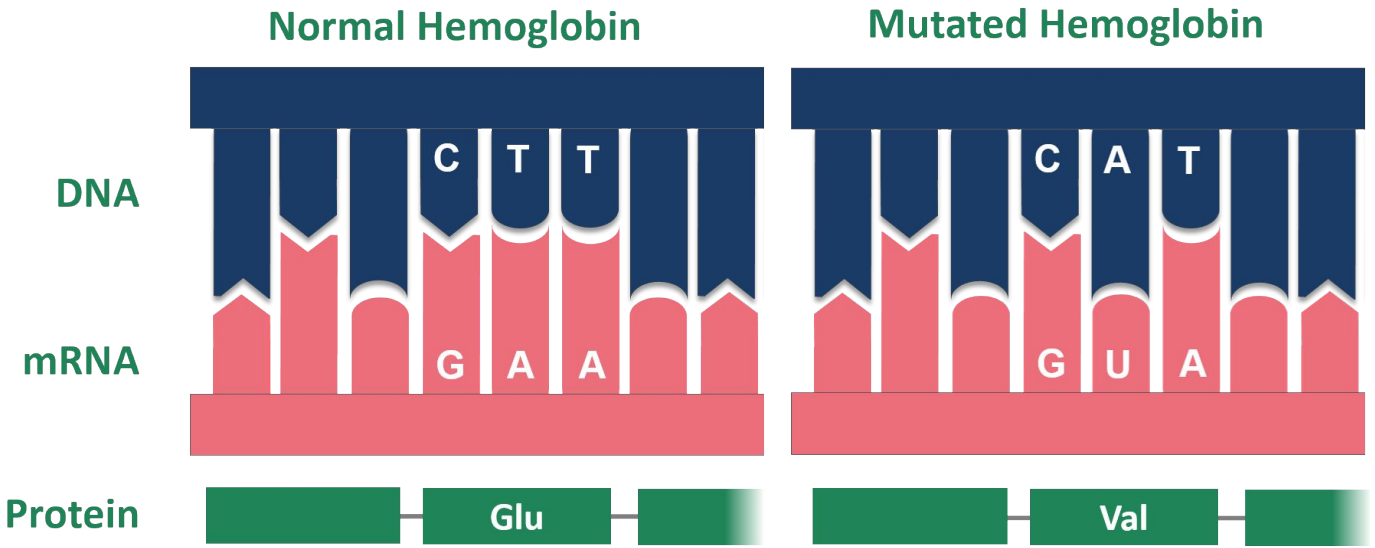


Sickle Cell Anemia (SCA)

- Harmful and beneficial
- Disadvantage:
  - affects function of haemoglobin; rbc changes shape (C-shape) when exposed to low oxygen levels
- Advantages
  - Protects against malaria

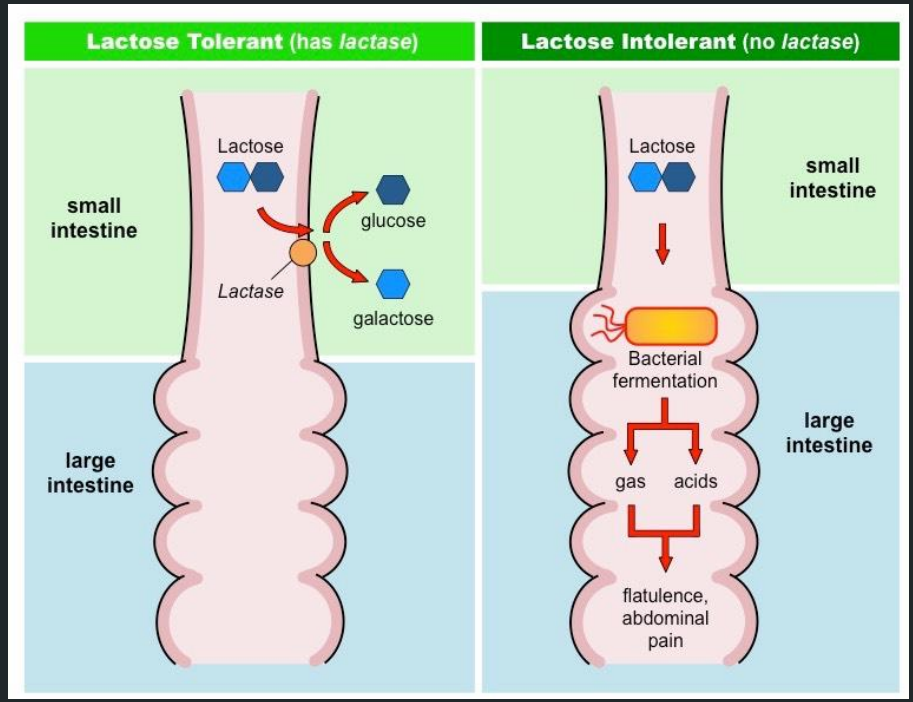
Sickle Cell Anemia - Substitution **Point** Mutation

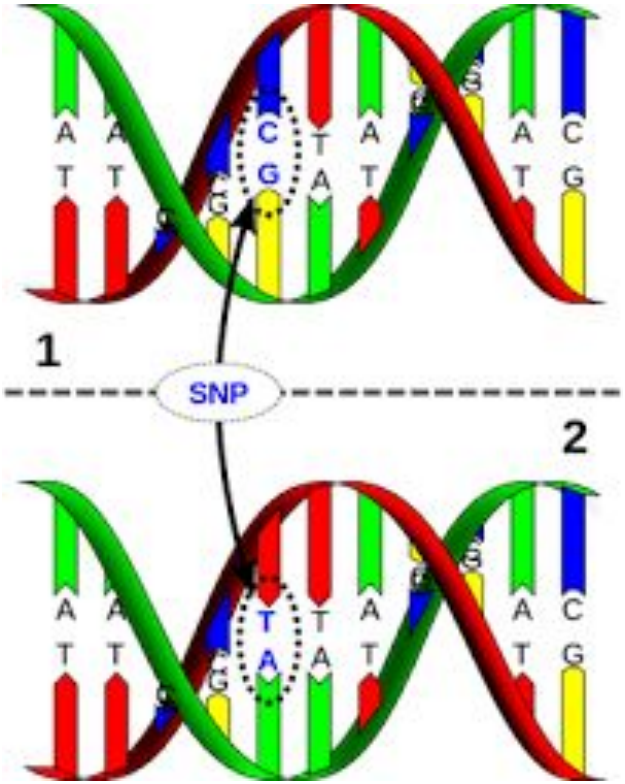
- Adenine nucleotide substituted for a **thymine nucleotide**
- Results in the production of altered **protein** (altered function)



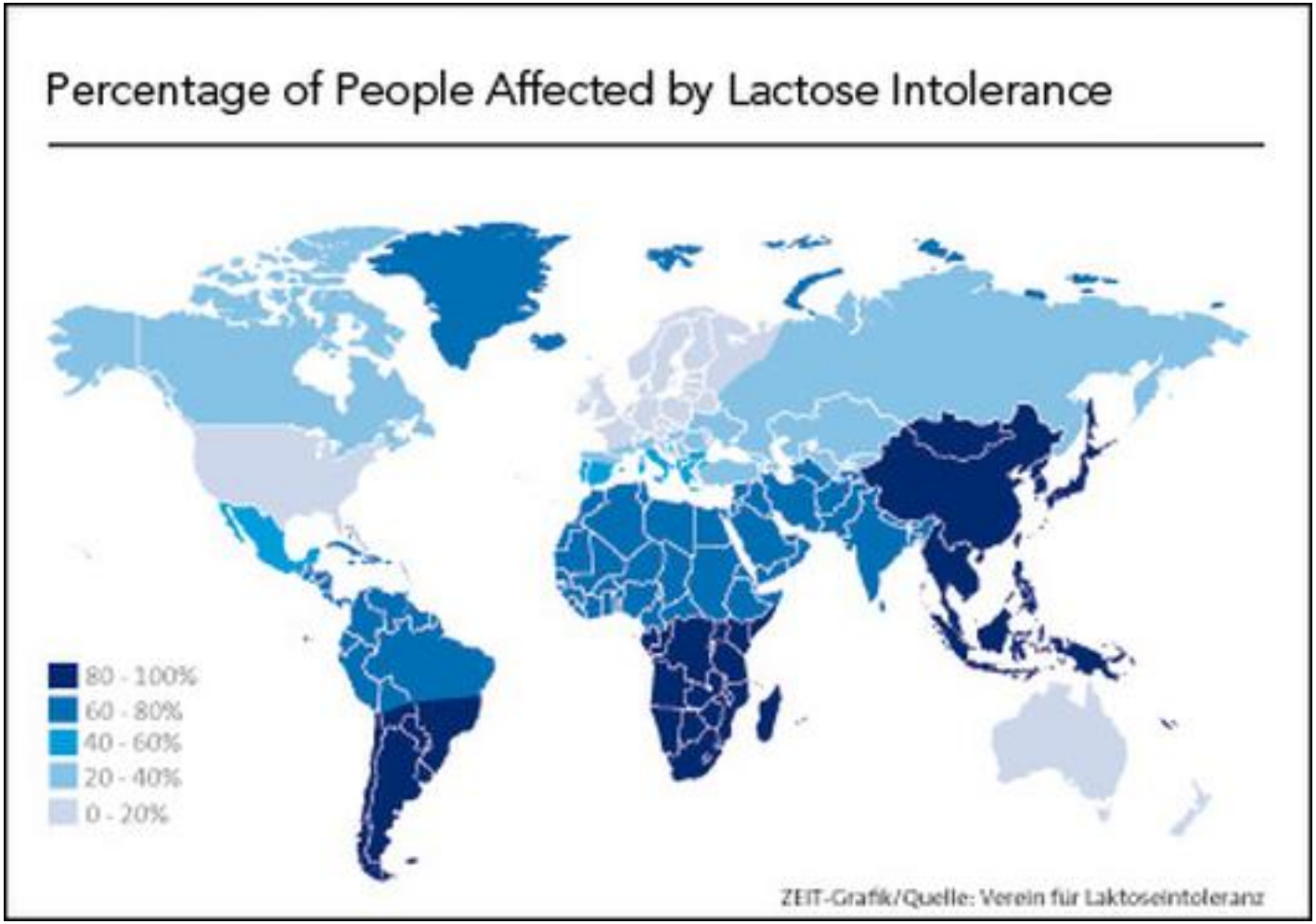
Lactose Intolerance & Tolerance

- People who are lactose intolerant do not produce enzymes (lactase) to break down lactose





- Most people (75%) are lactose intolerant worldwide
- Lactose **tolerant** people have a **mutation in chromosome 2!**
- Genetic tests can help you determine if you are lactose intolerant


Percentage of People Affected by Lactose Intolerance
80 - 100%
60 - 80%
40 - 60%
20 - 40%
0 - 20%
ZEIT-Grafik/Quelle: Verein für Laktoseintoleranz

Mutations - **spontaneous** or **induced**?

Spontaneous Mutation

- A mutation that occurs in nature or by accident (randomly)
- A result of a copy error in replication for mitosis or meiosis

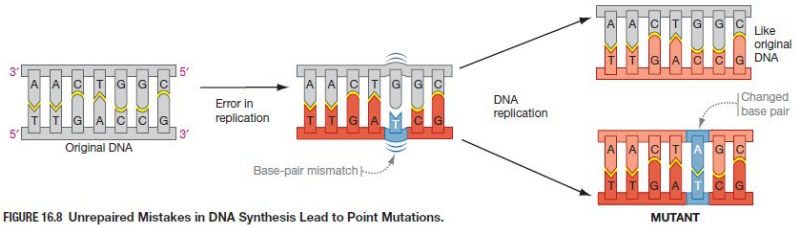


FIGURE 16.8 Unrepaired Mistakes in DNA Synthesis Lead to Point Mutations.

Induced Mutation

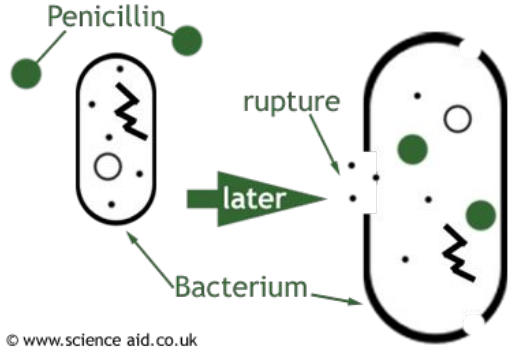
- A mutation that occurs because of exposure to an outside factor/ mutagen
- Examples:
  - Radiation
  - Cigarette smoke

Antibiotic Resistance

Antibiotic resistance:

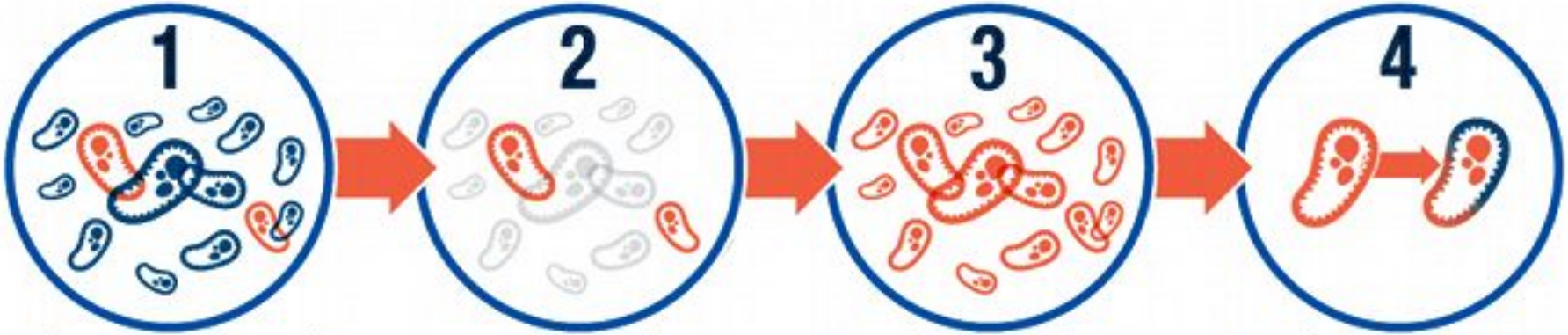
- describes strains of bacteria (superbugs) that are no longer susceptible to the effects of antibiotics

Normal mechanism of antibiotics: attach to specific bacterial cell wall causing bacterium's contents to burst out - bacteria dies.



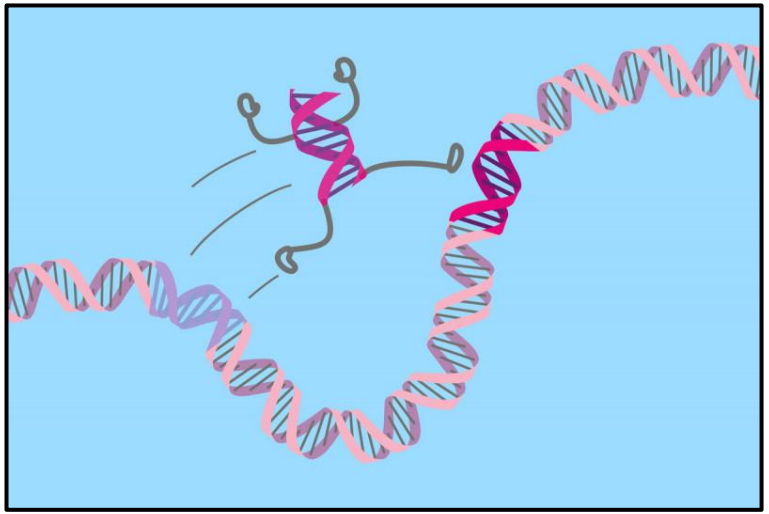
- **Problem:** a mutation occurs in the bacterial DNA that helps prevent destruction of cell wall. Resistant genes may be passed down.

How does antibiotic resistance occur?



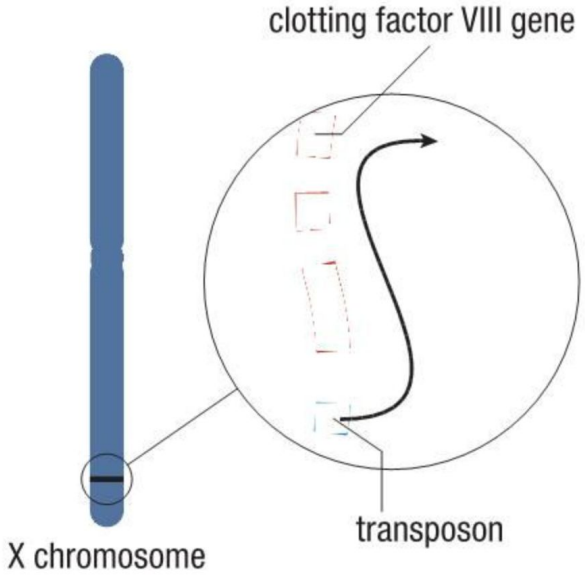
Jumping Genes

- Barbara McClintock discovered that an organism's genome is not static.
- **Transposon:** a specific segment of DNA that can move along or between chromosomes.
- **Transposition:** the process of moving a gene sequence from one part of the chromosome to another.



Hemophilia

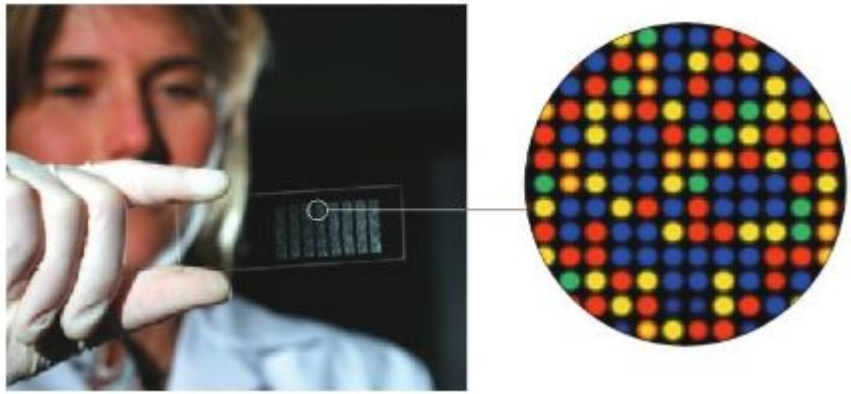
Jumping Genes



- Previously learned: hemophilia is an X-linked genetic disorder
- In rare cases, it can also be caused by a transposon if it inserts itself into a normal blood factor VIII gene
  - **Males:** one X chromosome has to be affected
  - **Females:** two X chromosomes have to be affected

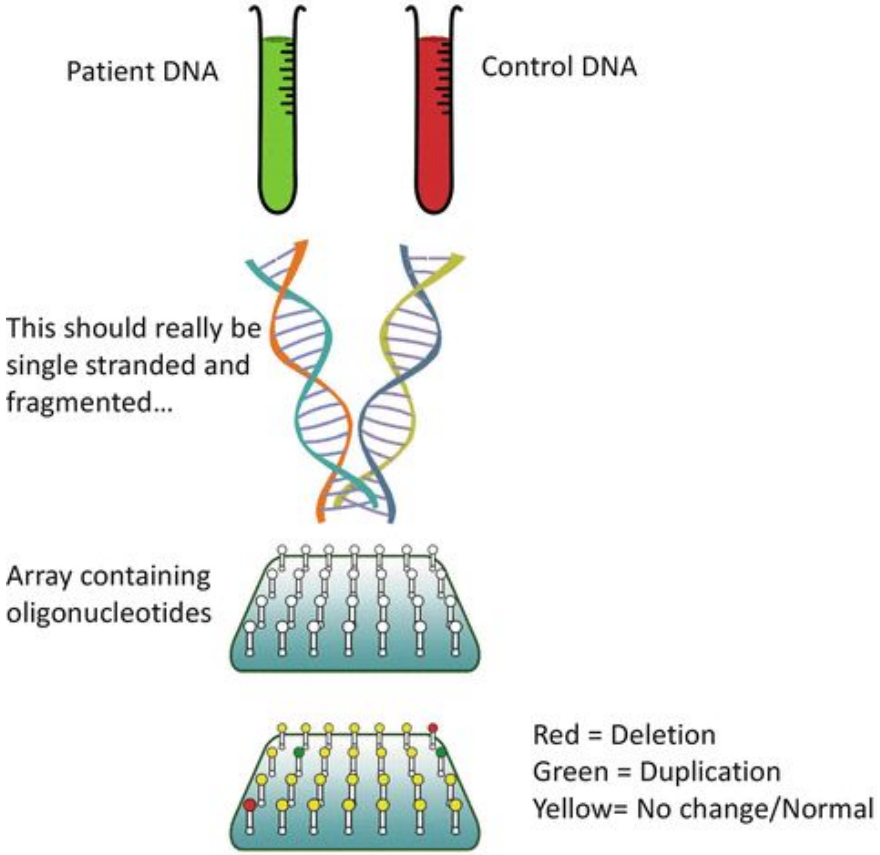
Microarray

Genetic Testing



- **Microarray:** a small membrane or glass slide that has been coated in a predictable and organized manner with a genomic sequence

- Can test hundreds or thousands of DNA fragments and determine if an individual has a specific genetic disorder.



Principle:

complementary sequences will bind to each other indicating presence of disease

6.2 Summary

6.2 Summary

- A mutation is change in the genetic code of an allele.
- Examples of point mutations are base-pair substitutions, insertions, and deletions. All result in a different protein being built by ribosomes.
- Lactose intolerance is the inability to digest lactose. Conversely, lactose tolerance is the ability to digest lactose. Lactose tolerance is the result of a mutation.
- Bacteria have developed antibiotic resistance due to mutations including those in the gene that directs the shapes of cell wall building-block molecules.
- Transposons are sequences of DNA that can move along and between chromosomes, possibly causing a change in an organism's phenotype.

Homework

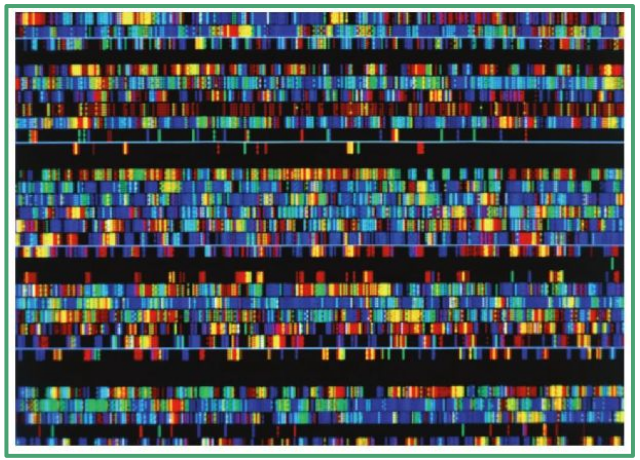
- Page 239 #2-5

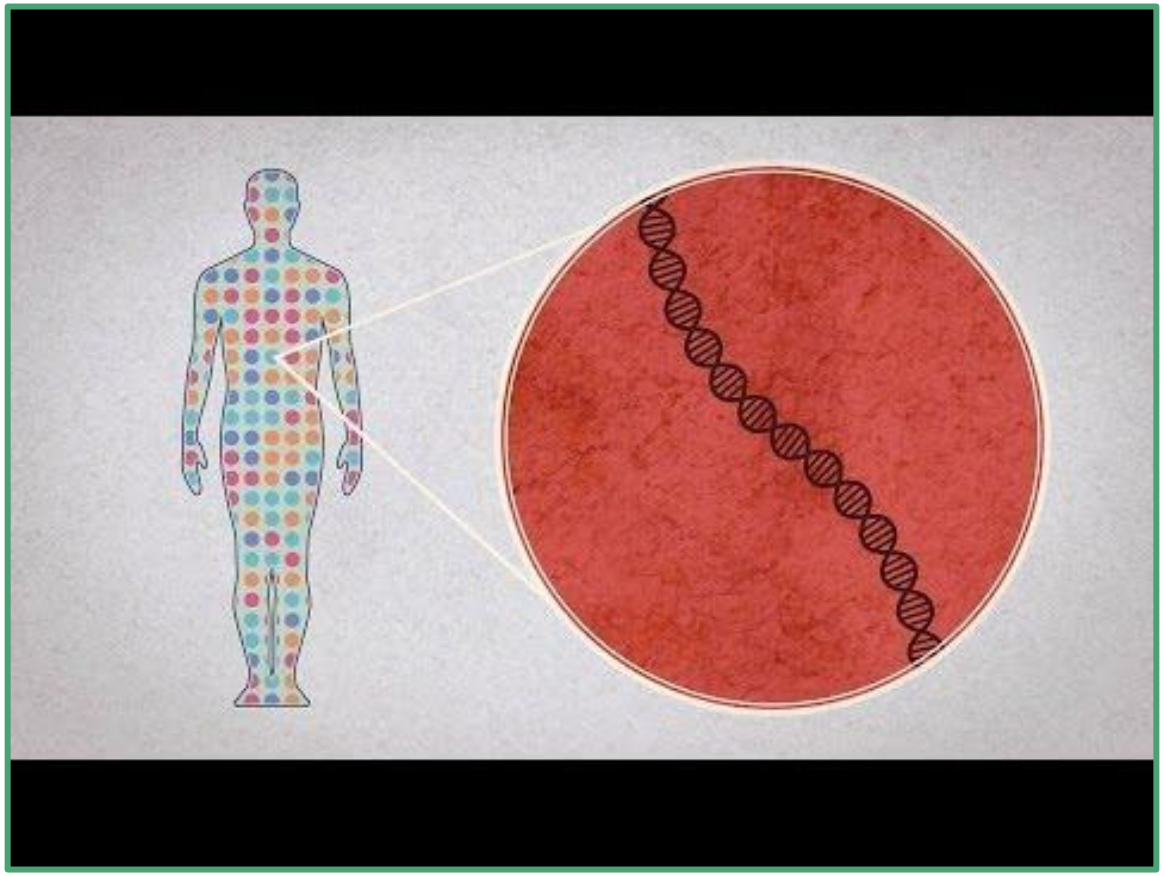
6.3 Genomes

SBI3U

The Human Genome Project

- There are 20,000 to 25,000 genes in the human genome
- Human DNA base sequences (order of A, C, T, G) have been analyzed
  - 3 billion base pairs in our DNA
- The order of the bases offers lots of information
- Scientists hope to use this information to cure diseases





Human Genome

Human Genome: the sequence of DNA nitrogenous bases found on the 23 sets of chromosomes in humans

The human genome is composed of both **coding** and **non-coding DNA**

Coding DNA (2% of human genome)

- Regions of the DNA that code for genes and proteins

Non-coding DNA (98% of human genome)

- Regions that do not code for proteins
    - The majority is considered junk DNA
    - Mutations here are not a concern

Other Genomes

Comparison of numbers of genes

Table 1 Number of Genes Believed Present in Different Genomes

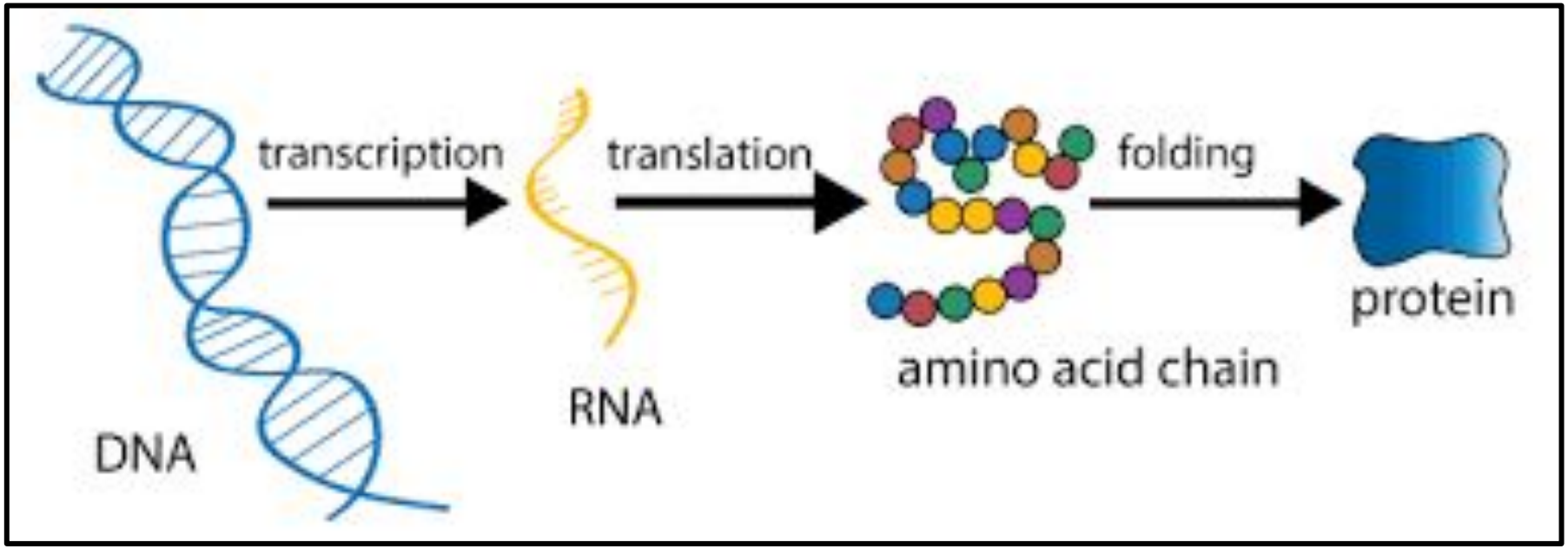
Organism	Size of genome	Approximate number of genes
amoeba	670 billion	unknown
newt (salamander)	84 billion	unknown
wheat	17 billion	unknown
human	3 billion	20 000
mouse	2.6 billion	25 000
Asian rice	446 million	50 000
fruit fly	137 million	13 602
yeast	12.1 million	6 034
E. coli	4.6 million	3 200
influenza virus	1.8 million	1 700

The water **amoeboid** has the largest genome followed by the newt (salamander).

- This does not necessarily correspond to number of **coding** genes.

Functional Genomics

- After determining the sequence of the genome, scientists wanted to learn about the relationship between genes and their function.
- Information can be used to screen for diseases and to help make medical decisions

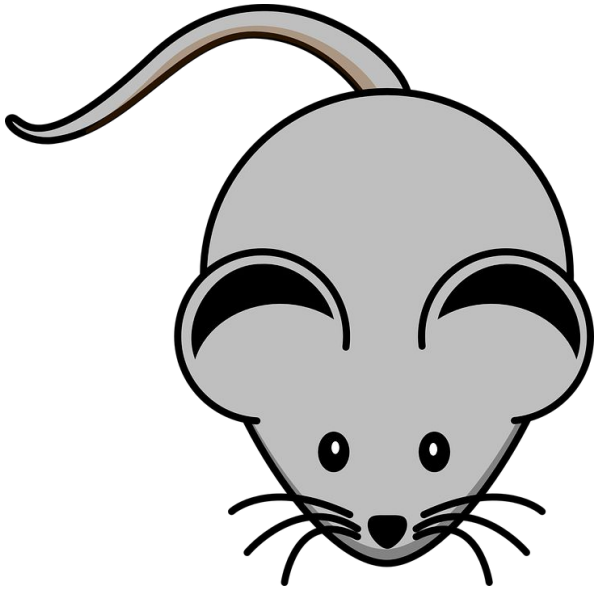


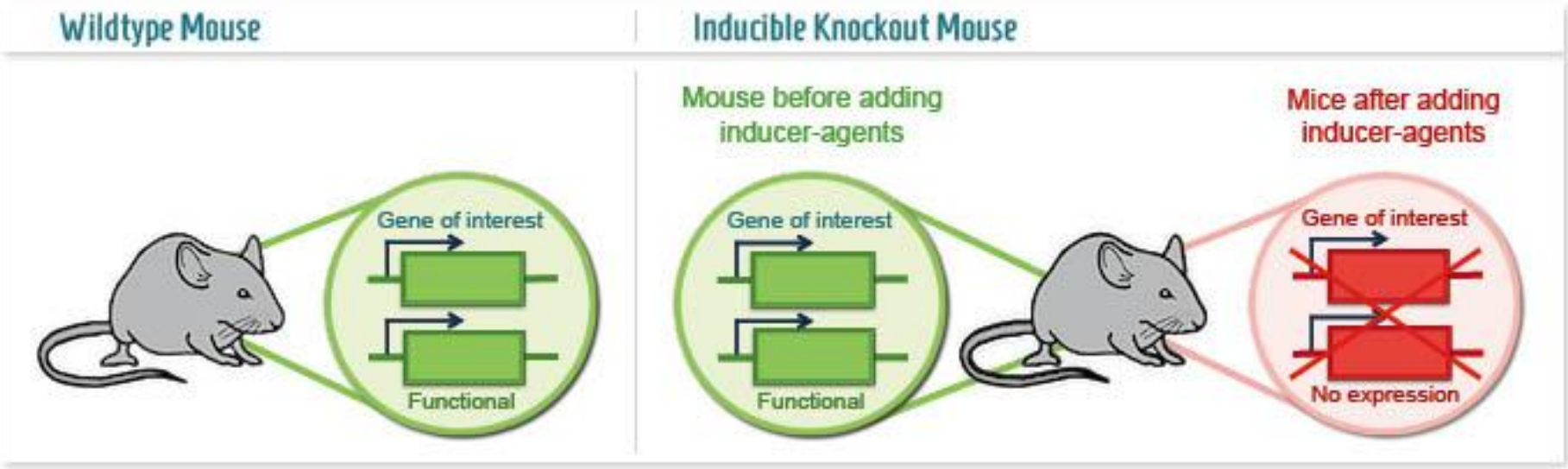
Functional genomics is used to determine the function or roles of genes in an organism.

Functional Genomics - Model Organism

- **Model organism:**
  an organism that can be used to study biological functions of another organism due to genetic similarity.

- **Mice** are model organisms for humans
  - Similarities between numbers and types of genes
    - E.g. both have gene that codes for hemoglobin (some phenotypic differences)





By mutating specific genes (in knockout mice), scientists can, prevent the formation of normal proteins, and observe how the loss of the gene affects the mice. This can in turn give scientists information on the normal function of the gene.

Knockout mice

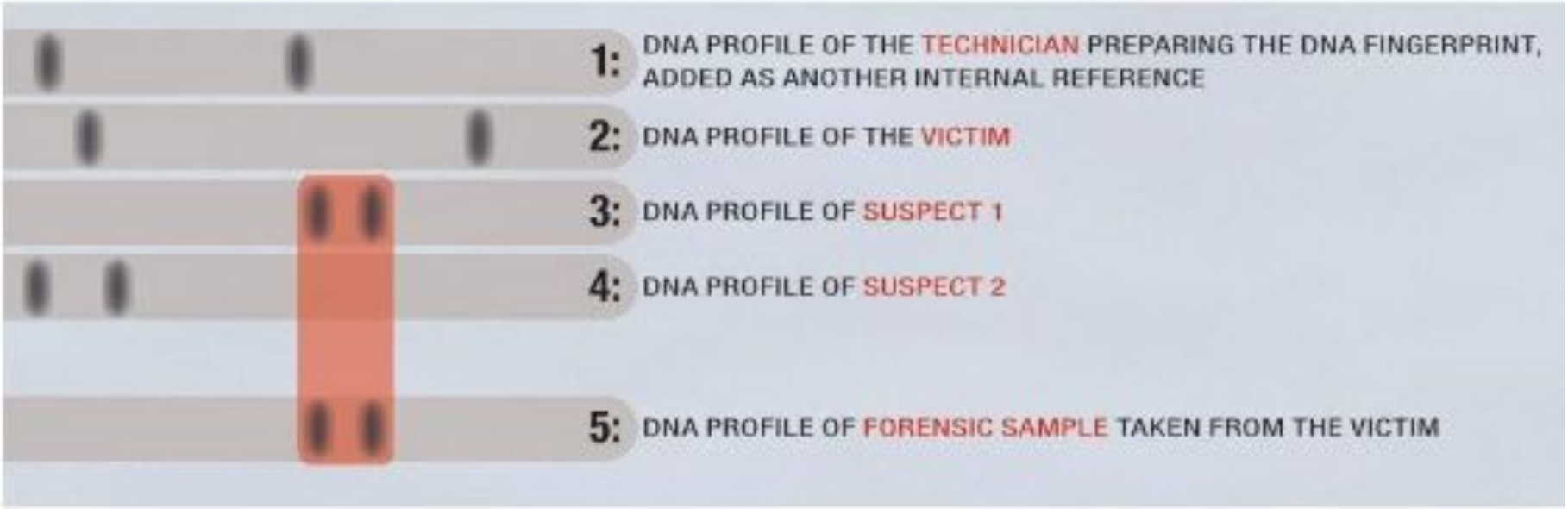
Figure 3 There is a 95 % to 98 % similarity between the genomes of humans and apes.

DNA Banks

- DNA information is considered personal (unless under investigation)
- A DNA bank is a database of DNA sequences (stored samples of DNA); the sequences can be from plants, animals or humans
- Can be used for screening and genetic testing
- Can be used to store DNA of endangered species

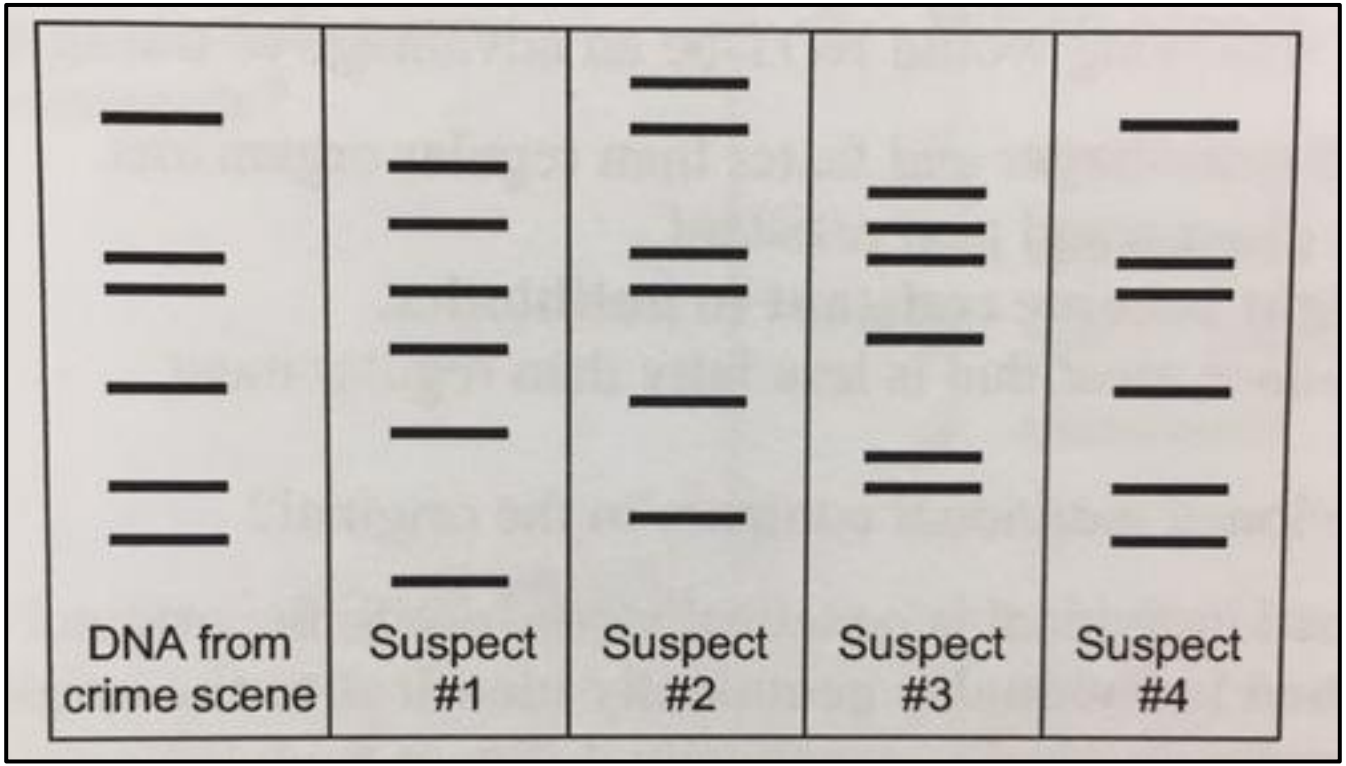
DNA Fingerprinting

- a technique that creates an image/pattern of bands of DNA on a gel that is unique to each individual



Which suspect matches the DNA fingerprint sample?

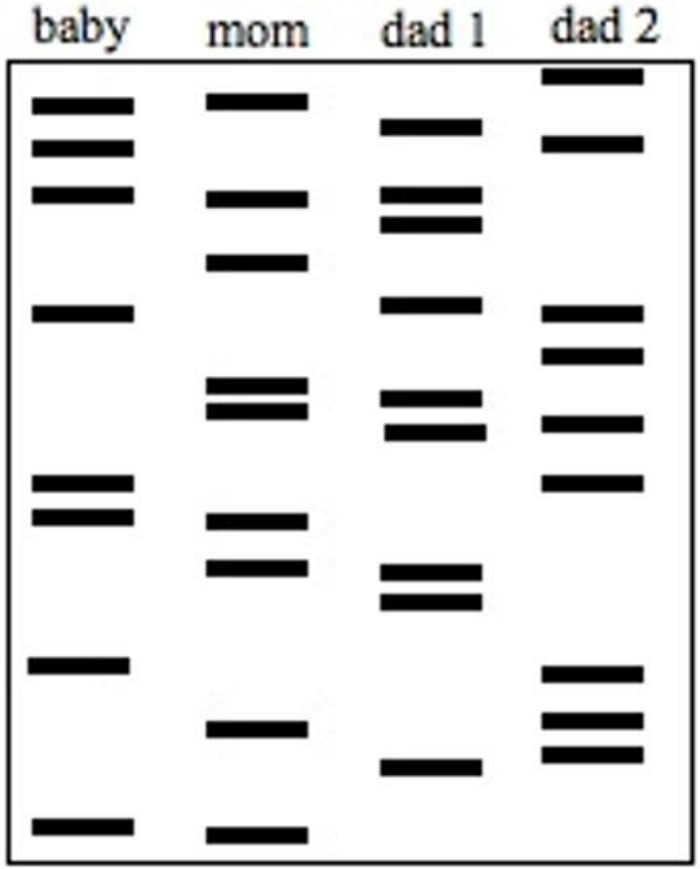
Note: *forensic detectives perform many tests and compare many bands from each sample before determining likelihood of a match*



Who is the offender?

DNA Fingerprinting - Uses

- To prove guilt or innocence
- Paternity tests
- Determining ancestry
- Identifying individuals in unmarked graves
- To prosecute hunters or fishers who hunt or fish certain animals out of season
- To determine true source of animal meat



6.3 Summary

6.3 Summary

- The Human Genome Project and Celera Genomics successfully sequenced the 3 billion base pairs in the human genome.
- Different genomes contain different numbers of genes. The human genome contains approximately 20 000 genes.
- The mouse genome is very similar to the human genome in gene function. Mice are good model organisms for human genetic studies.
- DNA banks can be used to store DNA for future use. DNA data banks also store the DNA of endangered species and plants, as well as fish.
- DNA fingerprinting is a technique that may link a suspect to a crime scene.
- Forensic DNA evidence is frequently used in Canadian criminal trials and even in fish and wildlife enforcement.
- The National DNA Data Bank in Canada stores the DNA of convicted criminals.

Homework

- Page 244 #1-6 & 8