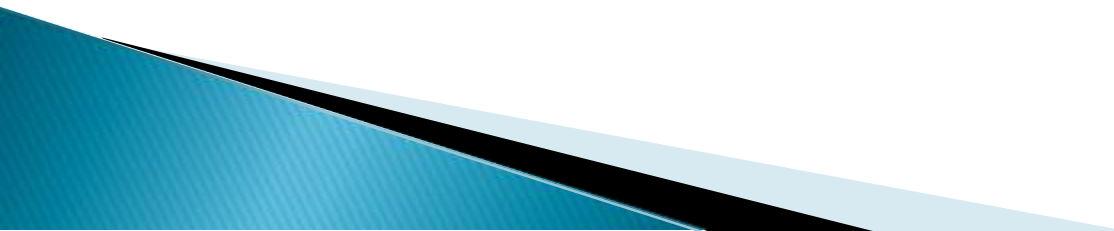


**Variations and Mutations
instead of PPT USE
BIOLPGY STUDY GUIDE**

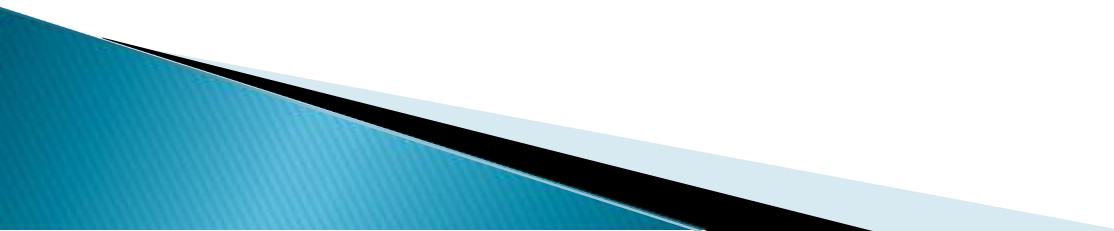


Lesson Objectives

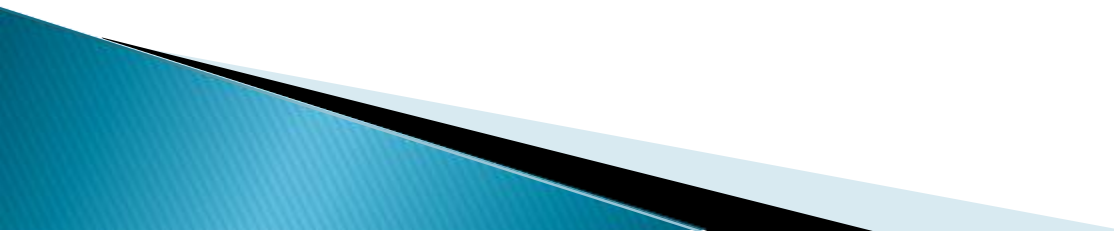
At the end of this lesson you should be able to

1. Identify two causes of variation
 2. Identify the characteristics of mutant alleles
 3. Identify two types of mutations
 4. Give an example of each type
 5. List two agents responsible for increased mutation rates
- 

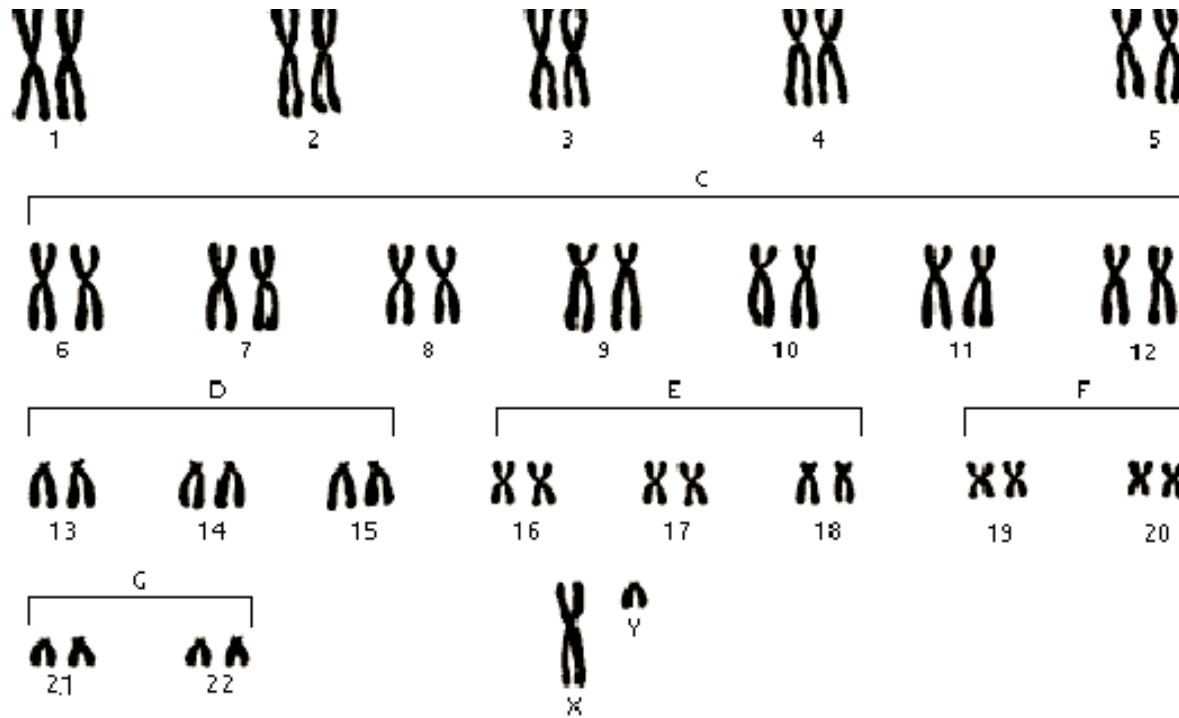
Chromosomes and Genetics

- ▶ Chromosomes are long pieces of DNA, with supporting proteins
 - ▶ Genes are short regions of this DNA that hold the information needed to build and maintain the body
 - ▶ Genes have fixed locations: each gene is in a particular place on a particular chromosome
 - ▶ Diploids have 2 copies of each chromosome, one from each parent. This means 2 copies of each gene.
- 

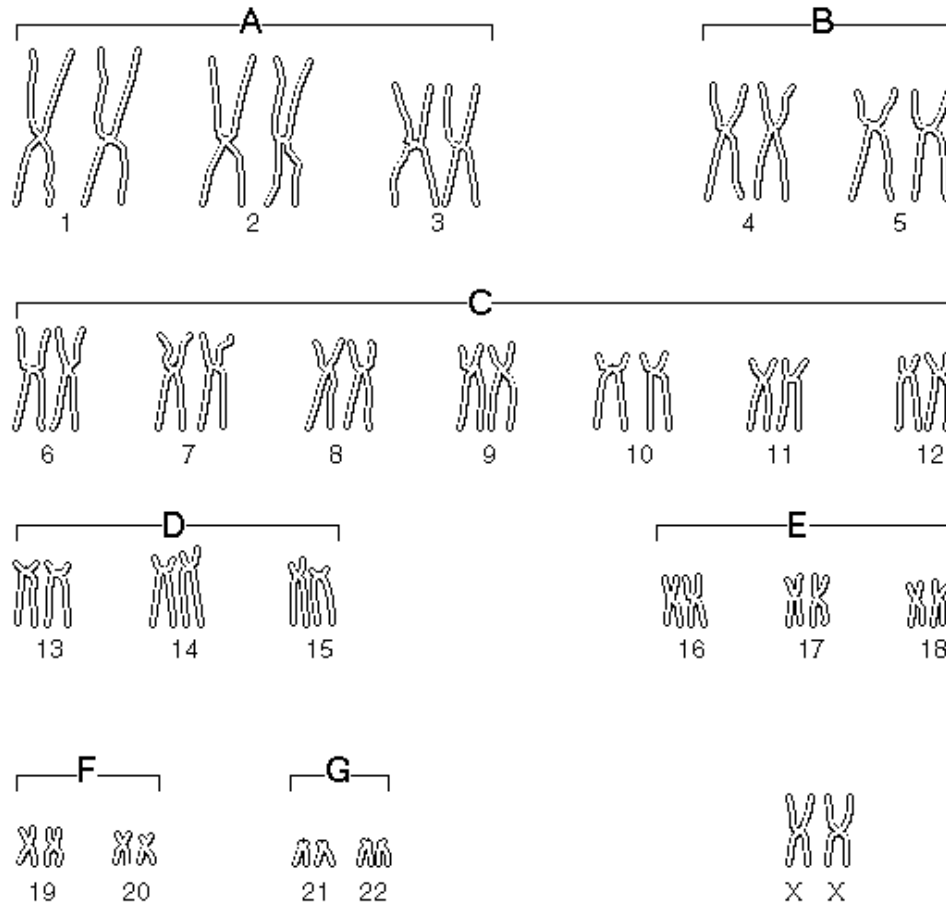
Human Chromosomes

- ▶ We have 46 chromosomes, or 23 pairs.
 - ▶ 44 of them are called autosomes and are numbered 1 through 22. Chromosome 1 is the longest, 22 is the shortest.
 - ▶ The other 2 chromosomes are the sex chromosomes: the X chromosome and the Y chromosome.
 - ▶ Males have an X and a Y; females have 2 X's: XY vs. XX.
- 

Male Karyotype



Female Karyotype



Sex Determination

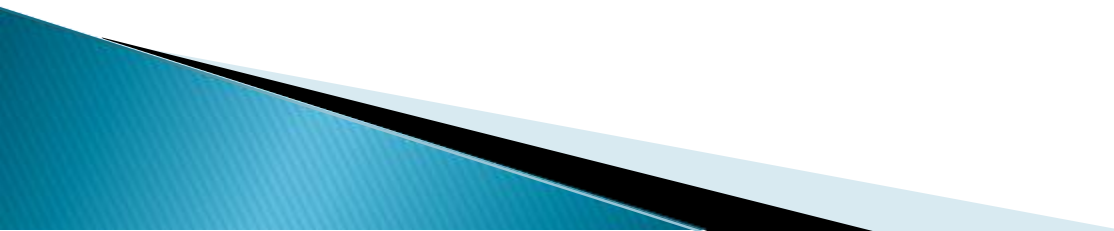
The basic rule:

If the **Y chromosome** is present, the person is **male**.

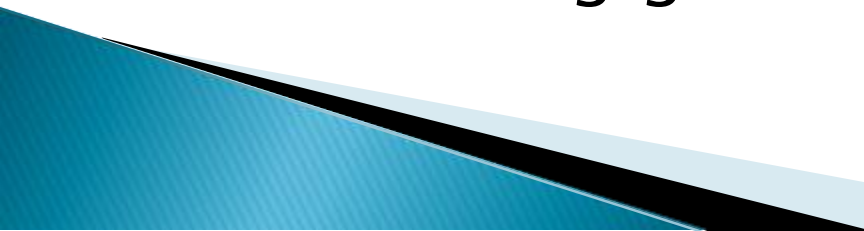
If **absent**, the person is **female**.



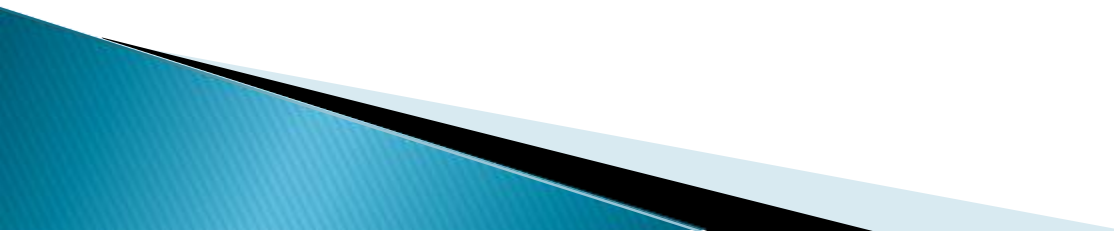
Learning Check

1. What is the difference between a chromosome and a gene?
 2. What is the difference between the terms haploid and diploid?
 3. How many pairs of chromosomes do we have?
 4. What are pair number 23 called?
 5. What is the difference between the male and the female karyotype?
- 


Meiosis

- ▶ the X and Y chromosomes separate and go into different sperm cells:
 - ▶ $\frac{1}{2}$ the sperm carry the X and the other half carry the Y.
 - ▶ All eggs have one of the mother's X chromosomes
 - ▶ The Y chromosome has the main sex-determining gene on it, called SRY
- 

Sex Determination

- ▶ About 4 weeks after fertilization, an embryo that contains the **SRY gene** develops testes, the primary male sex organ.
 - ▶ The testes secrete the hormone testosterone.
 - ▶ Testosterone signals the other cells of the embryo to develop in the male pattern.
- 

Causes of Inherited Variations

1. Sexual reproduction
 2. The independent assortment of chromosomes when gametes are being formed
 3. Crossing Over during Meiosis – this allows genes to move between chromosomes
 4. Mutations
- 

Fact

When your parents cells undergo meiosis to produce sperm/egg cells

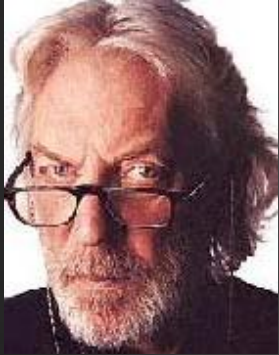
8 million different combinations of their chromosomes are possible

Every zygote is a 1 in 64,000,000,000,000 combination of chromosomes!

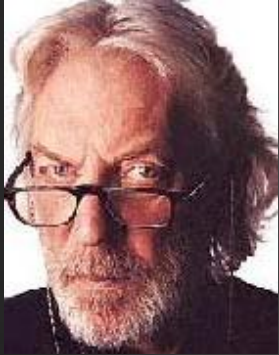
This leads to great variation in species



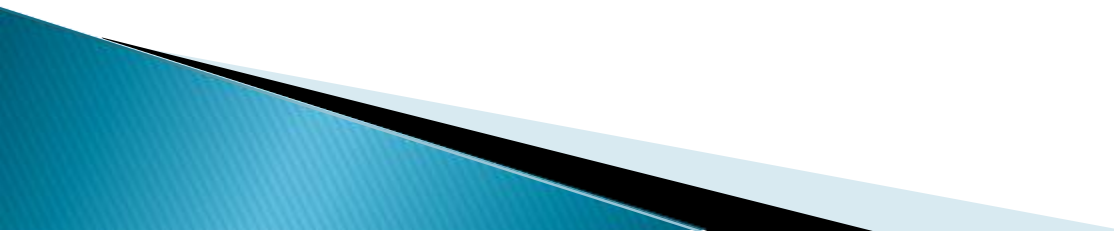
Whose Child?



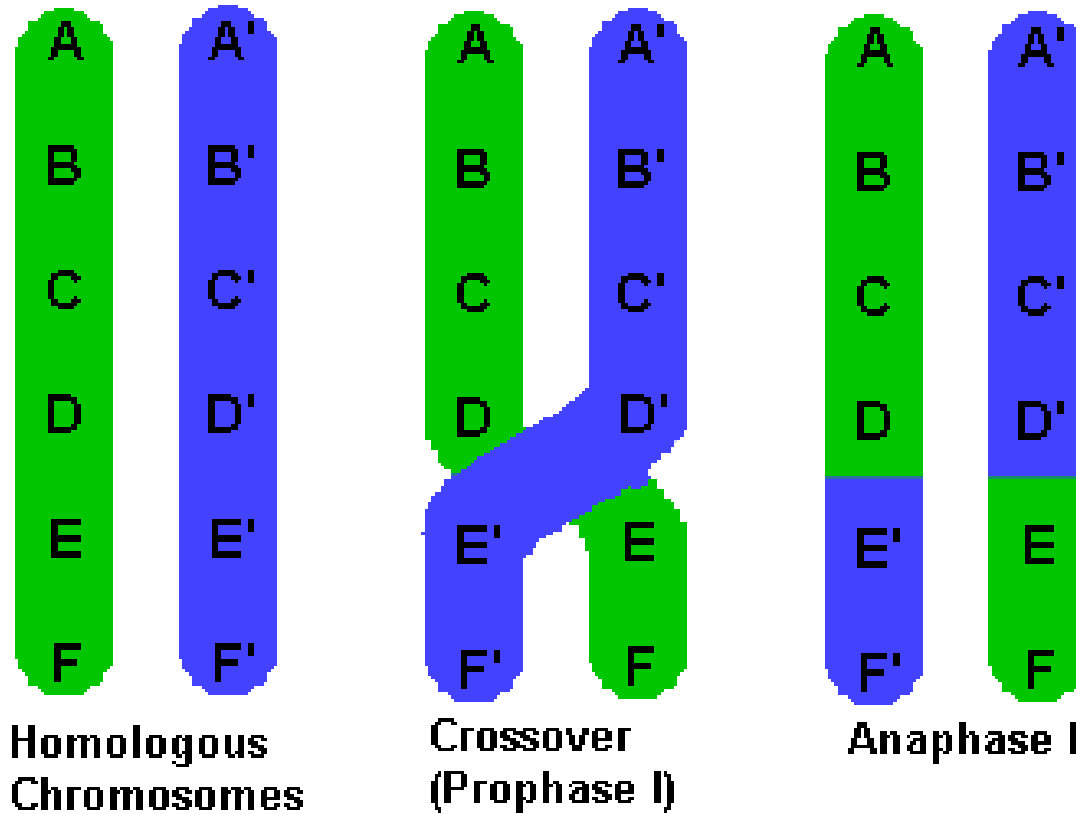
Whose Child?



Differences between parents and offspring

- ▶ Offspring are similar to their parents but are not identical
 - ▶ Cause – crossing over
- 

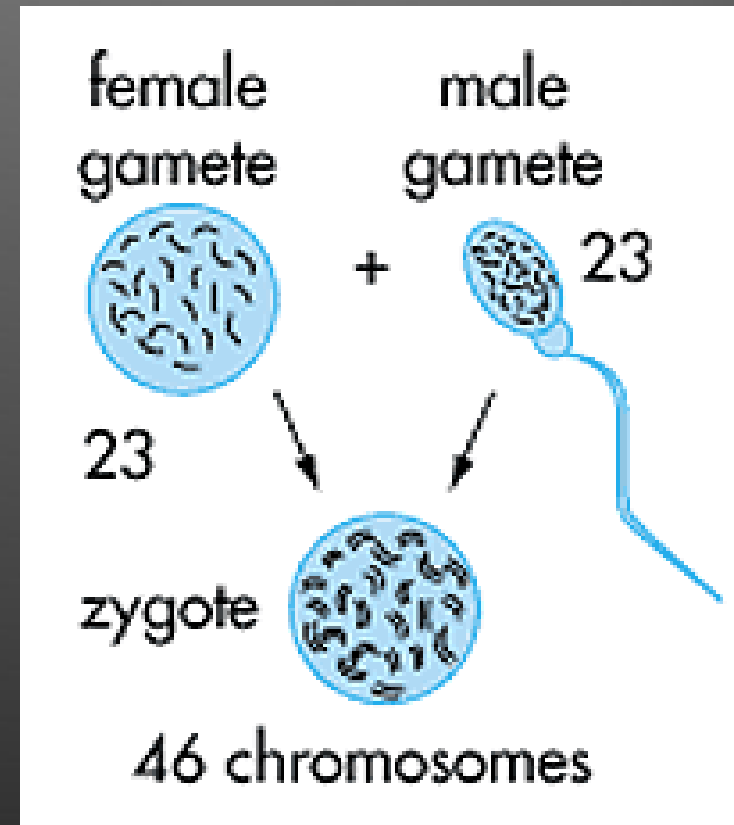
Crossing Over



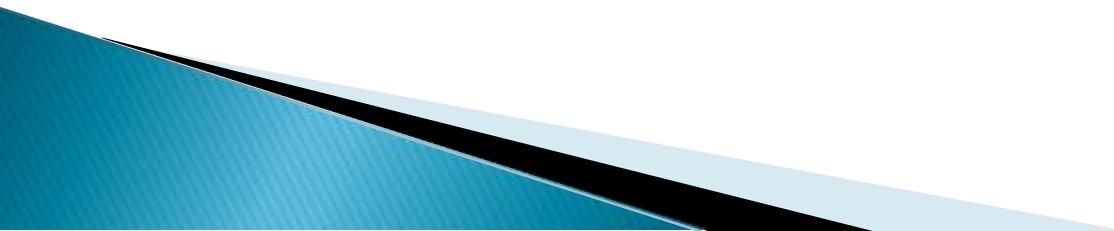
Fusion of gametes

Sperm and egg cell
fuse to form a
zygote


The combination of
chromosomes leads
to great variation in
species



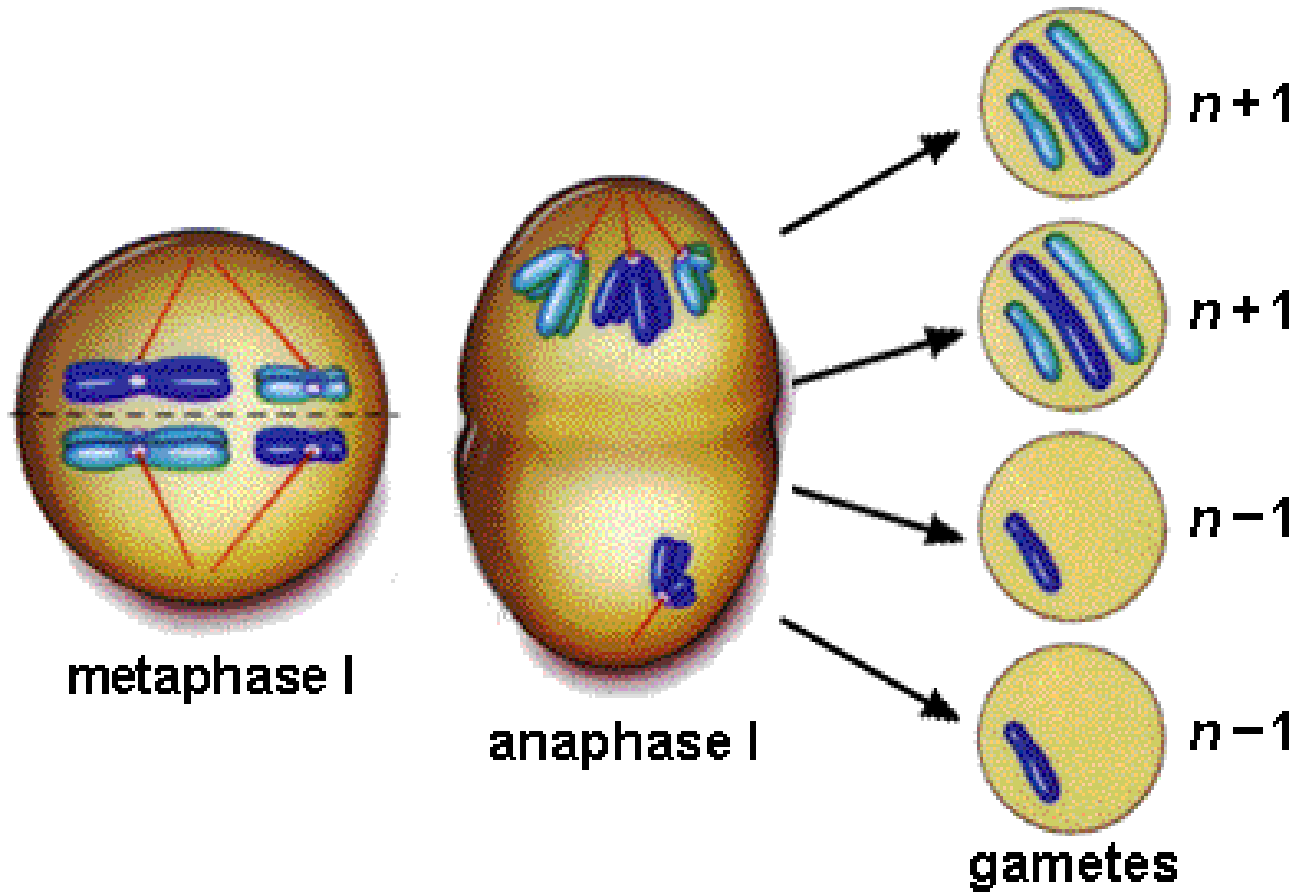
Learning Check

1. What happens during meiosis?
 2. What does meiosis lead to?
 3. What are the causes of variation?
 4. Why are offspring not identical to their parents?
- 

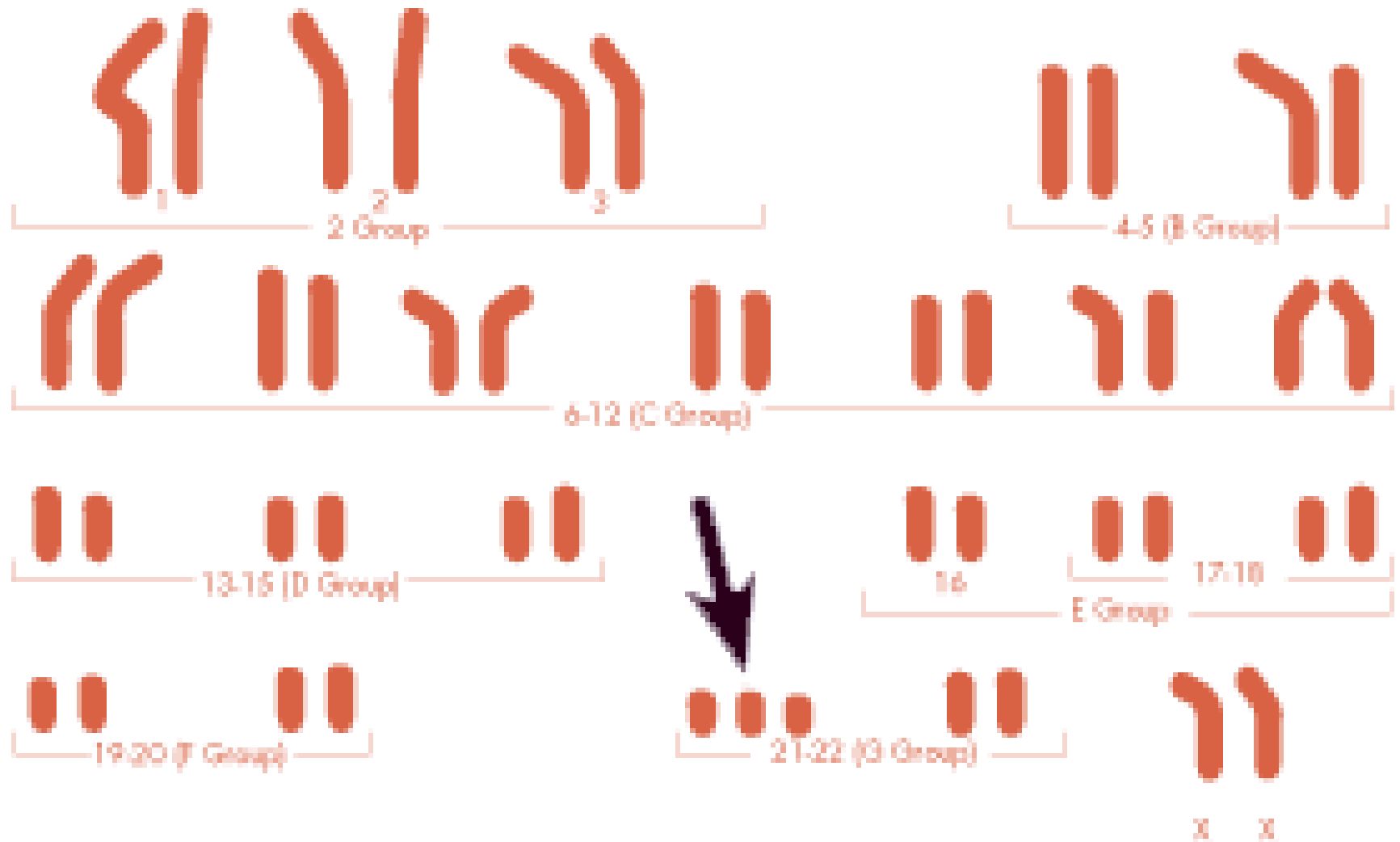
Chromosome Number Variations

- ▶ Except for the X and Y, humans don't survive with only 1 copy of any chromosome. Also, 3 copies is lethal in most cases.
 - ▶ Down Syndrome is the most common human aneuploidy. It is also called trisomy-21, meaning 3 copies of chromosome number 21.
- 

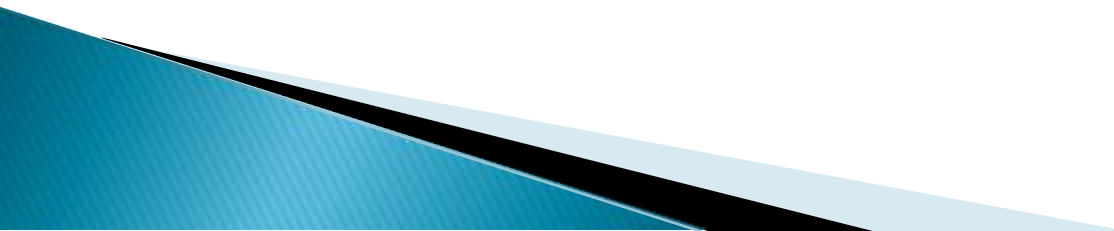
Aneuploidy



Downs syndrome

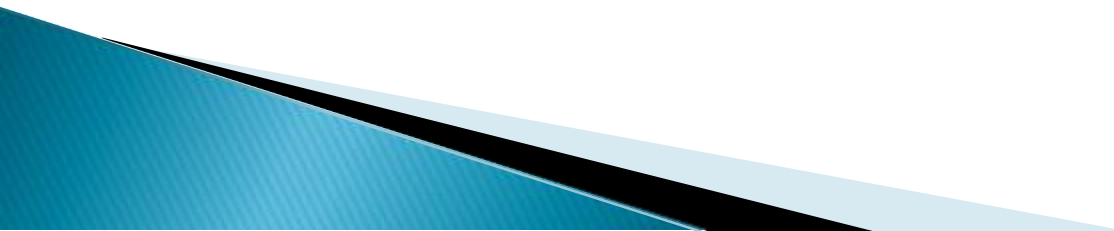


Chromosome Structure Variations

- ▶ Chromosomes can be broken by X-rays and by certain chemicals.
 - ▶ The broken ends spontaneously rejoin, but if there are multiple breaks, the ends join at random.
 - ▶ This leads to alterations in chromosome structure.
- 

Structure Variation Examples

There are lots of ways chromosomes can change structure, so the syndromes are not as well defined as with number variations.

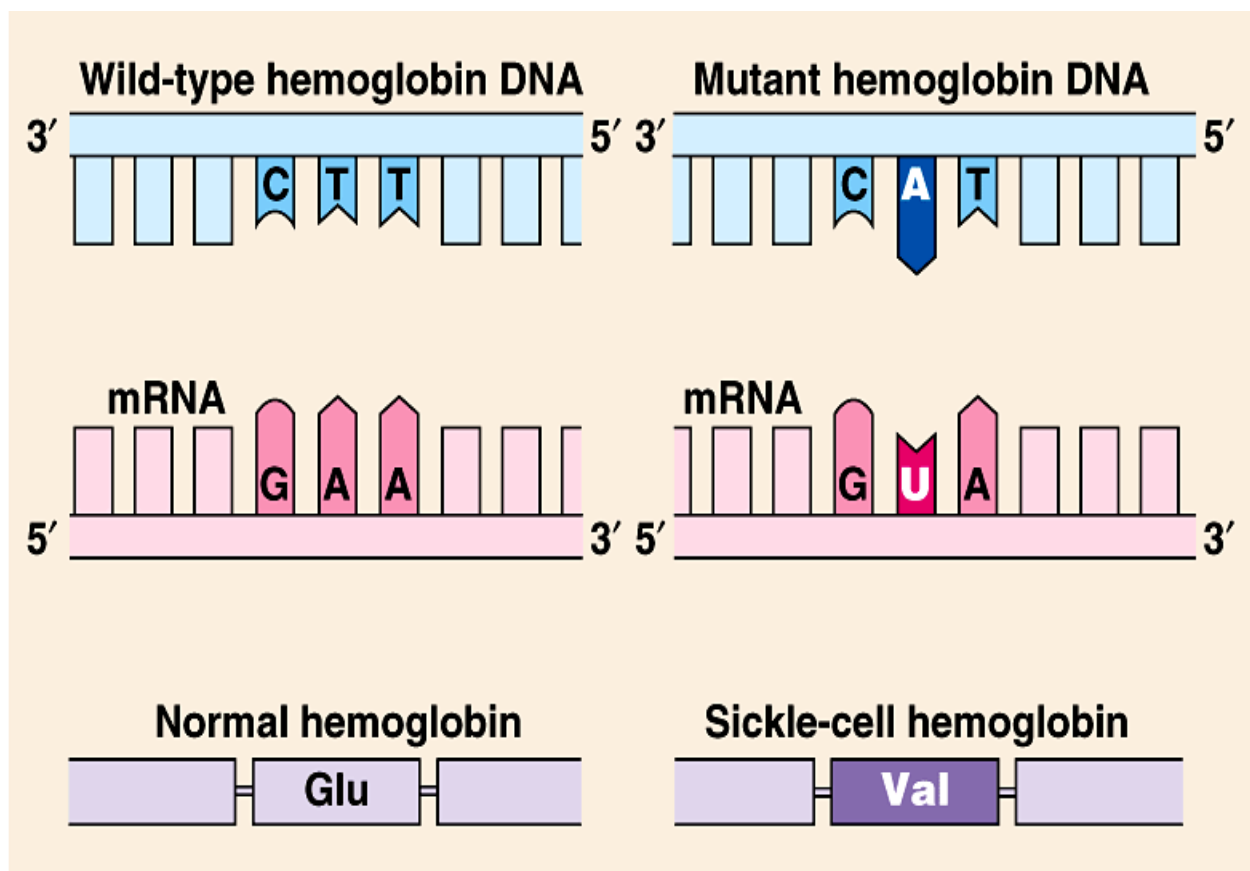


Point mutations

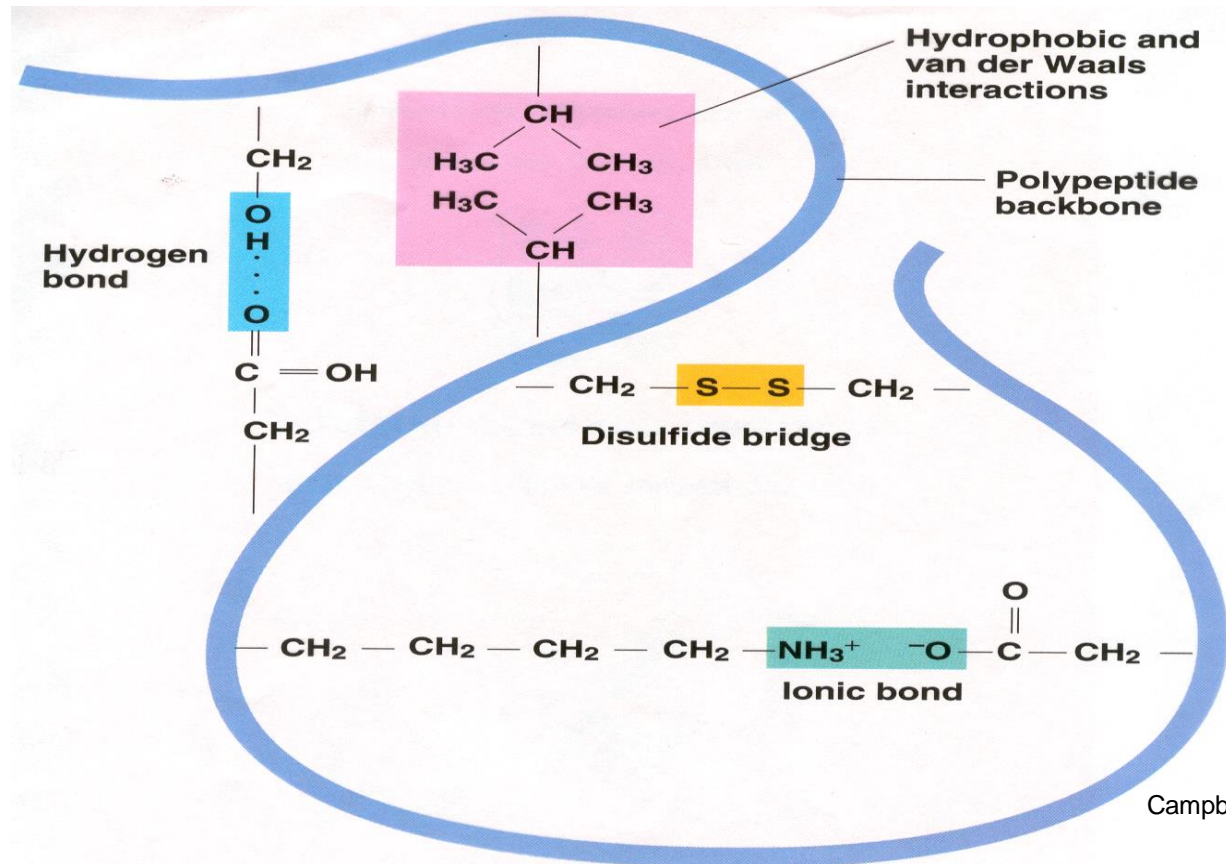
- ▶ These are changes in just one base pair of a gene.
- ▶ If a point mutation occurs in a gamete or in a cell that gives rise to gametes, it may be transmitted to offspring and to a succession of future generations e.g. sickle cell anaemia.

Sickle Cell Anaemia is caused by a point mutation-

A mutation of a single base pair in the gene that codes for one of the polypeptides of haemoglobin.



Protein folding



Campbell and Reece (2002) *Biology*

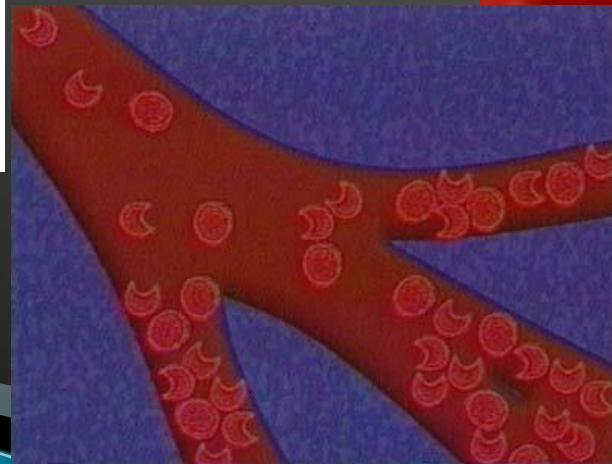
Just one amino acid could affect how a protein folds

As a result, damage is caused to the red blood cell membrane, resulting in a cellular lifespan of only a few days.

Due to the sickle cell shape, small capillaries may become blocked.



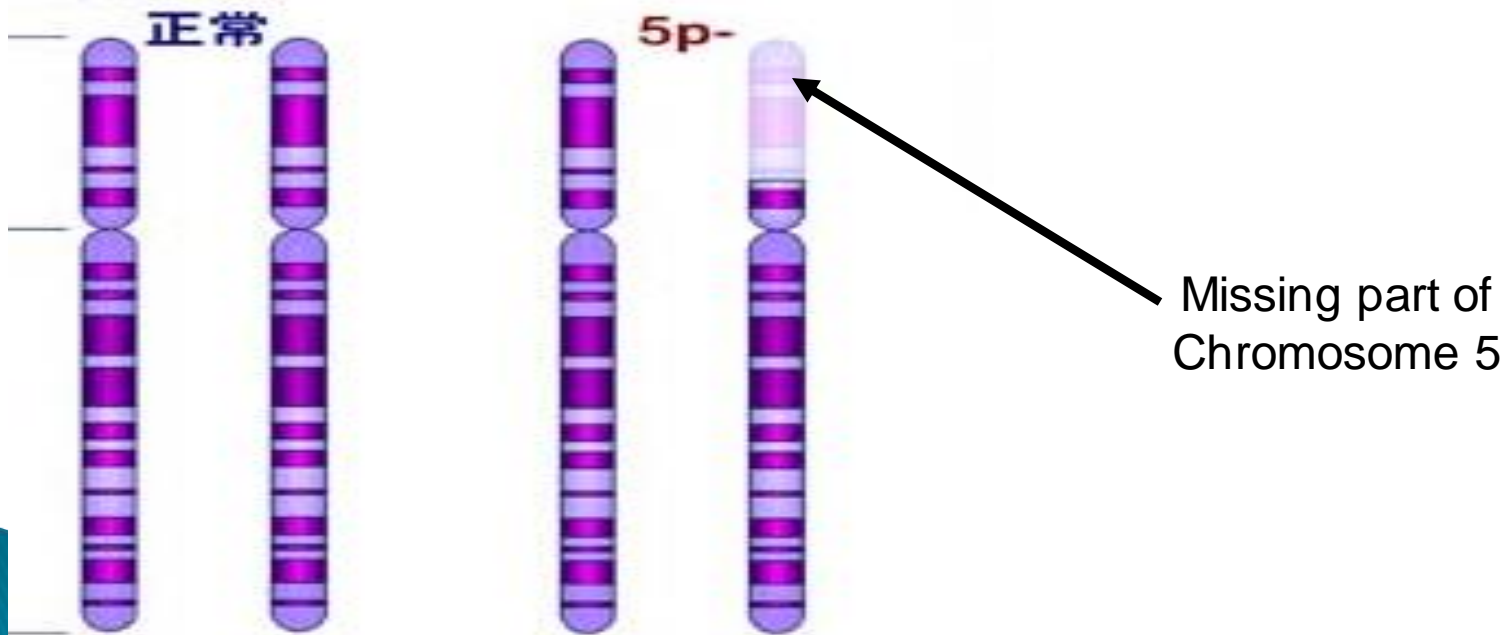
sciencemuseum.org.uk/genes/223.asp




www.pueblo.gsa.gov/sicklecell/496-sick.html

Cri Du Chat Syndrome

Cri-du-chat syndrome comes from a deletion of one end of chromosome 5, so the person only has 1 copy of all the genes on this end of the chromosome.



Mutations – Other Causes

- ▶ **Radiation** High energy radiation from a radioactive material or from X-rays
 - ▶ These X rays attack the DNA molecule and alters it in many ways.
 - ▶ Radiation can also cause double strand breaks in the DNA molecule, which the cell's repair mechanisms cannot put right
 - ▶ Remember Chernobyl
- 

Dying for a Suntan

Sunlight contains UV radiation (the component that causes a suntan)

This can cause an abnormal cross link in DNA to form between certain adjacent bases.

In most normal cases the cells can repair this damage, but sometimes a mutation can occur

Unprotected exposure to UV radiation by the human skin can lead to skin cancer and extensive skin tumors.

Smoking

- ▶ A chemical in cigarette smoke causes mutations in a crucial gene associated with cancer.
- ▶ Cigarette smoke caused mutations in a gene called P53.

Acetone _____

Mercury _____

Lead _____

Nicotine _____

Cadmium _____

Carbon Monoxide _____



_____ Hydrogen Cyanide

_____ Urethane

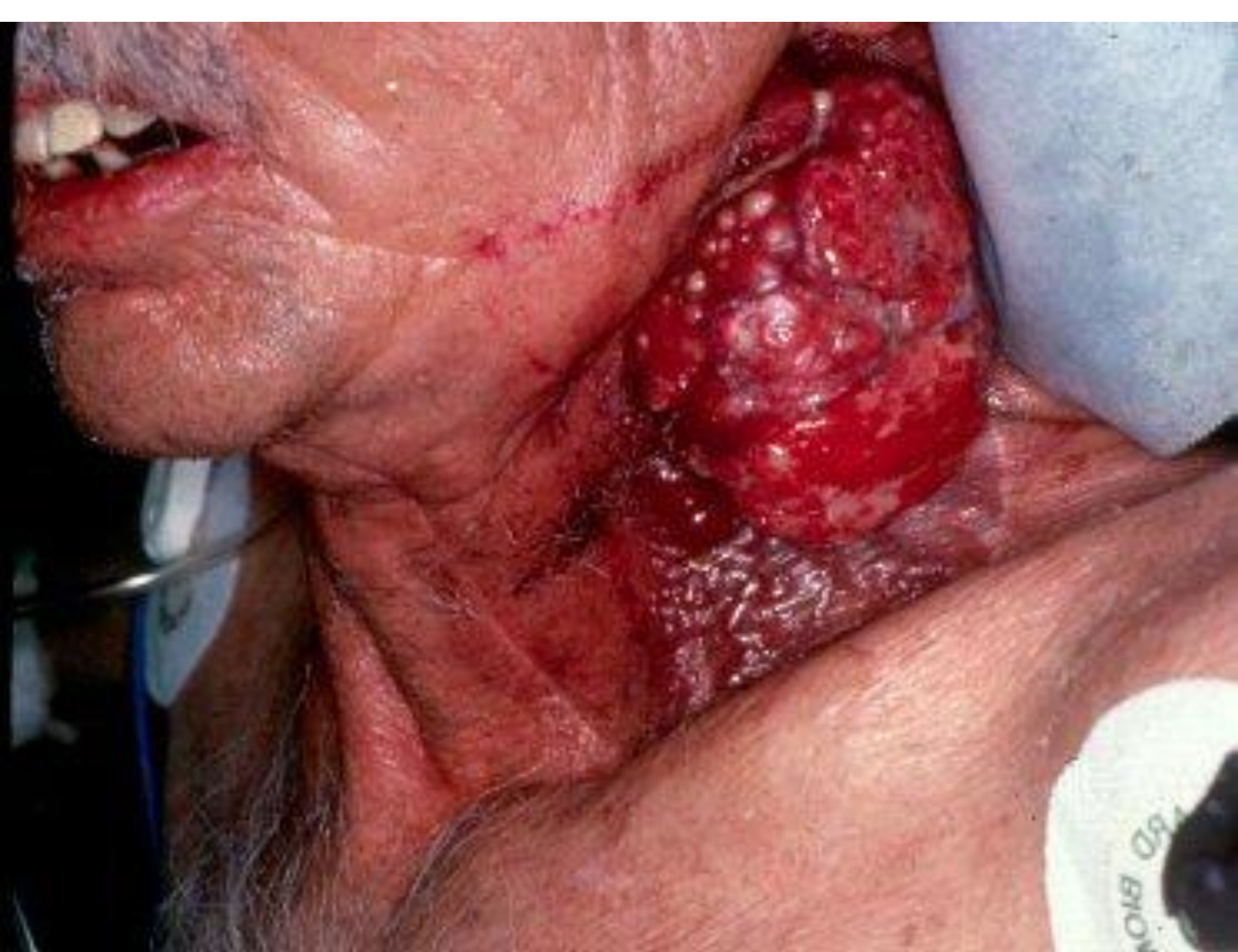
_____ Arsenic

_____ Phenol

_____ Formaldehyde

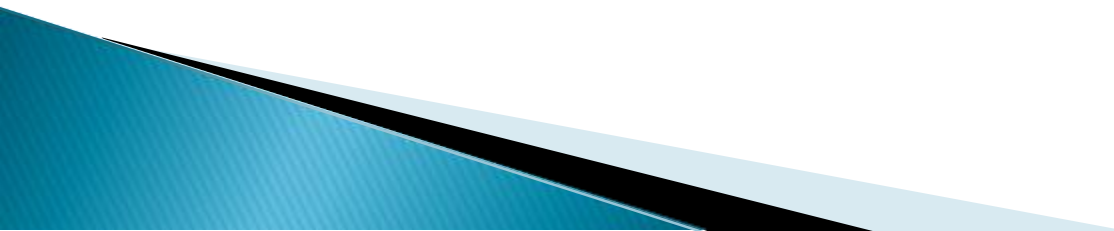
_____ DDT

Chemicals included in tobacco smoke



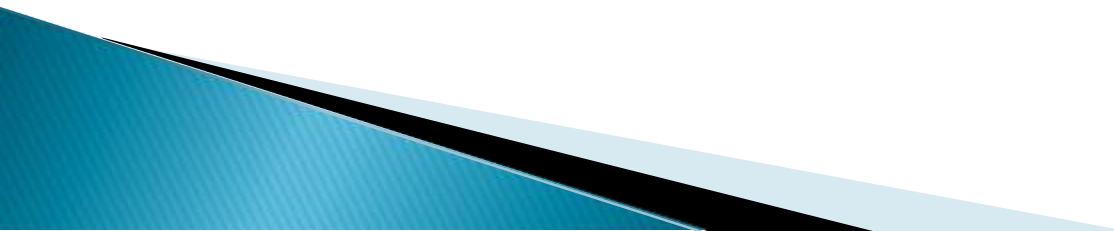


Learning Check

1. What is meant by aneuploidy?
 2. Give an example?
 3. What are the causes of structural variation in chromosomes?
 4. Give an example of a structural variation?
 5. What are the causes of mutations?
- 

What have you learned?

Can you

1. Define variation and mutation
 2. List the causes of variations and mutations
 3. List the types of mutations
 4. Discuss the causes for and effects of increased mutation rates
- 

End