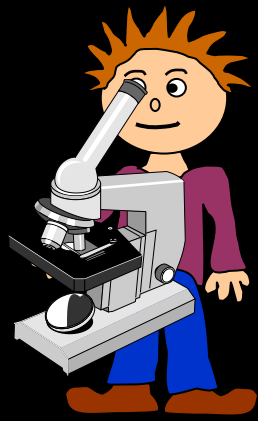
# You and your genes

(OCR)



# Variation

"Variation" is the name given to differences between individuals of the SAME species.

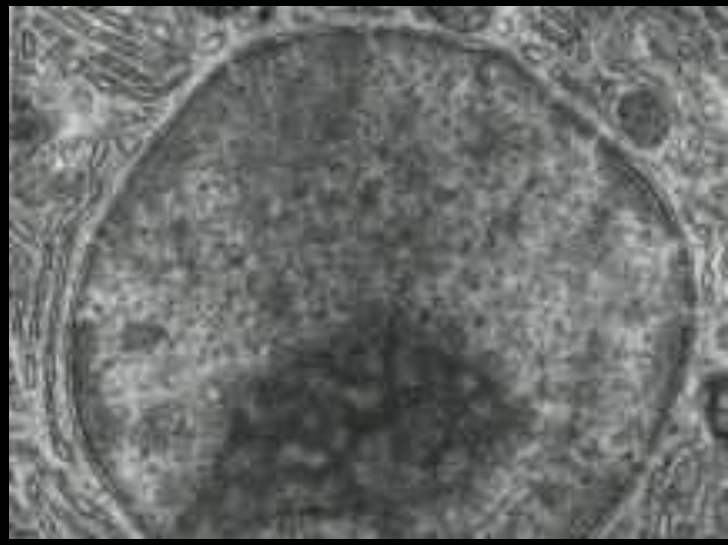
Variation is due to GENETIC or ENVIRONMENTAL causes. For example, consider dogs:

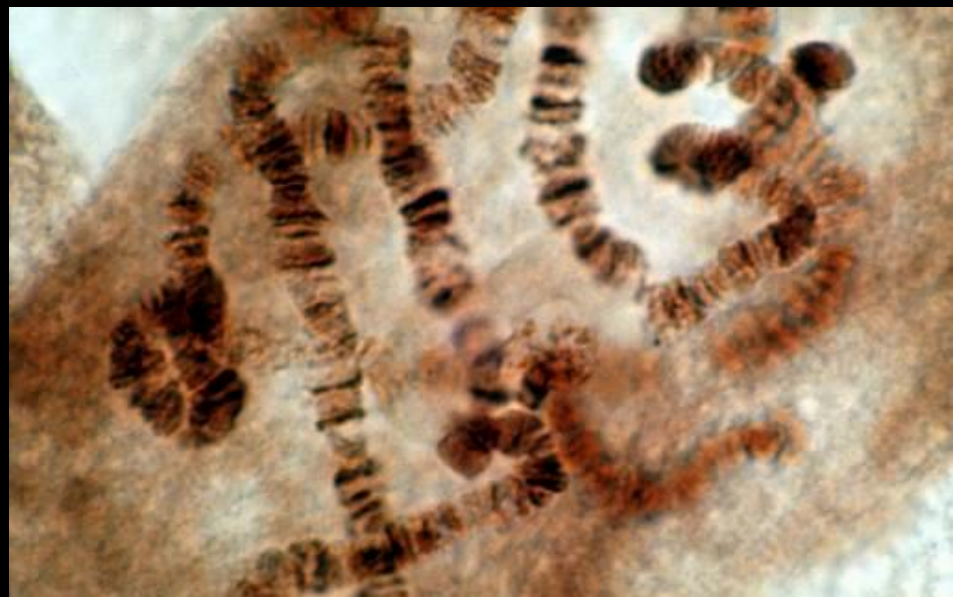
1) *Ways in which they are the same:*

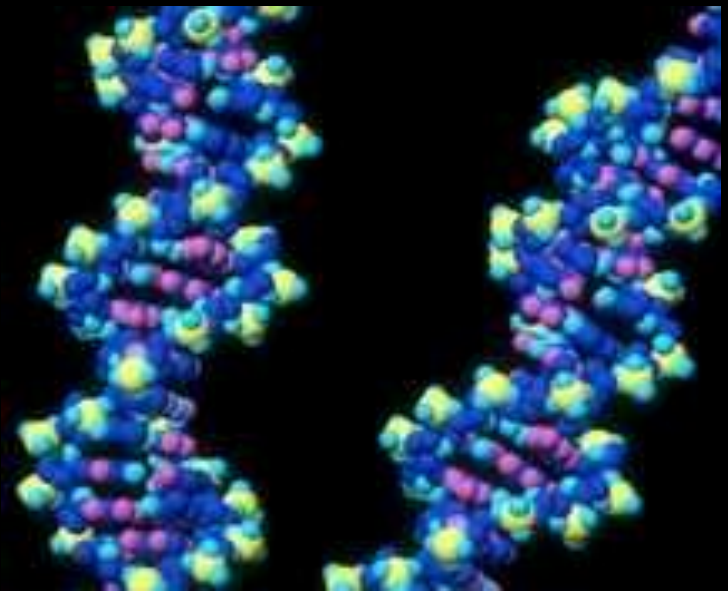
2) *Ways in which they are different:*

# Genes, Chromosomes and DNA

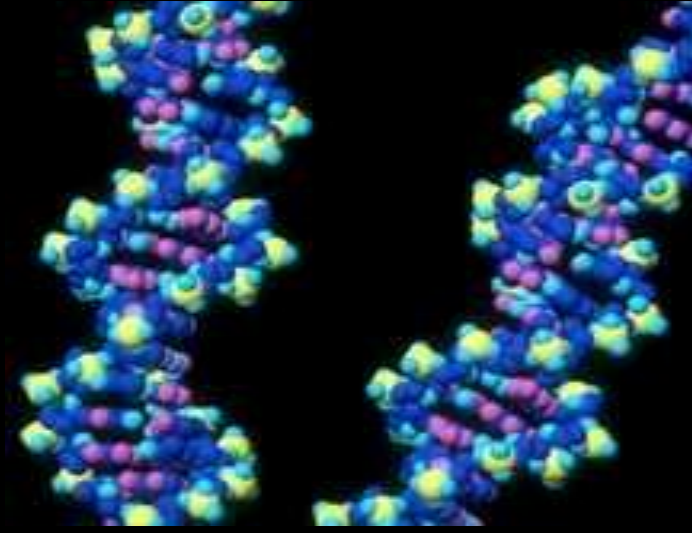
15/12/2011







# How genes work



**Some facts:**

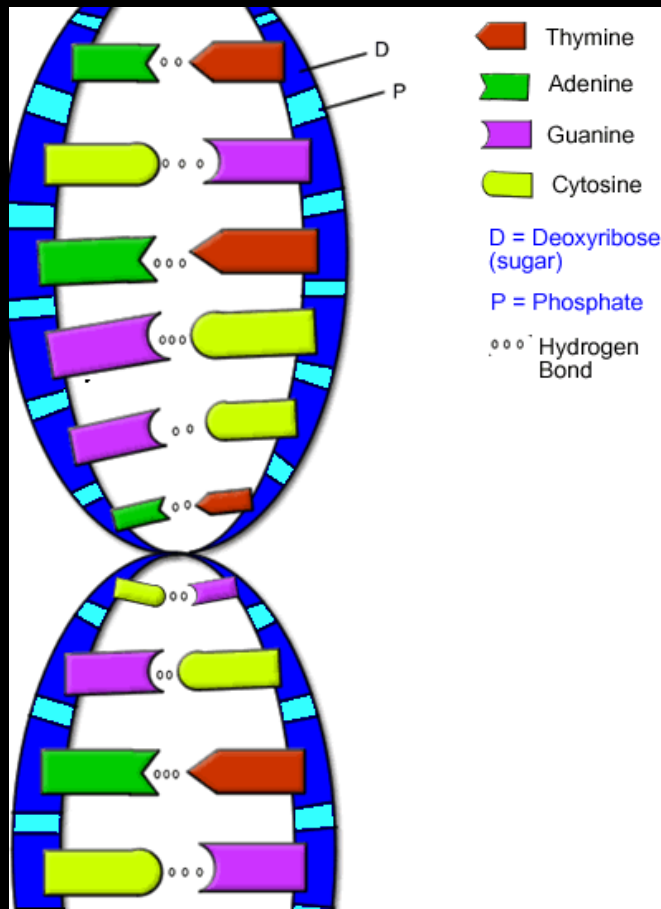
- The structure of DNA is called a "double helix"

- The DNA contains instructions on how the cell should work

- Genes control the development of characteristics ("it's in the genes") by issuing instructions to the cell to produce certain proteins

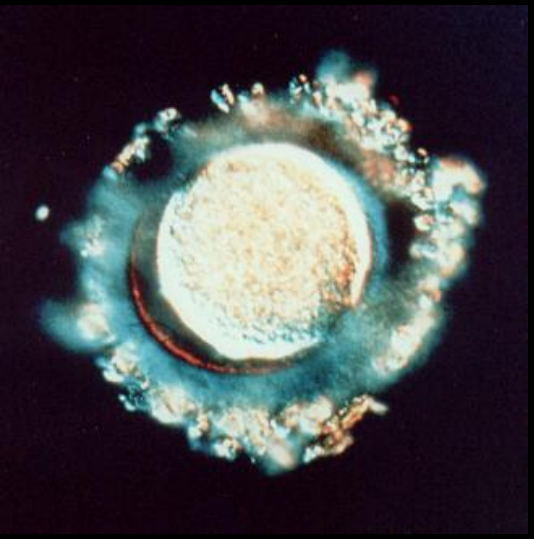
- These proteins are either structural (used for cell growth and repair) or enzymes (used for speeding up reactions)

- Genetic information can be transferred from one organism to another – this is "genetic modification"

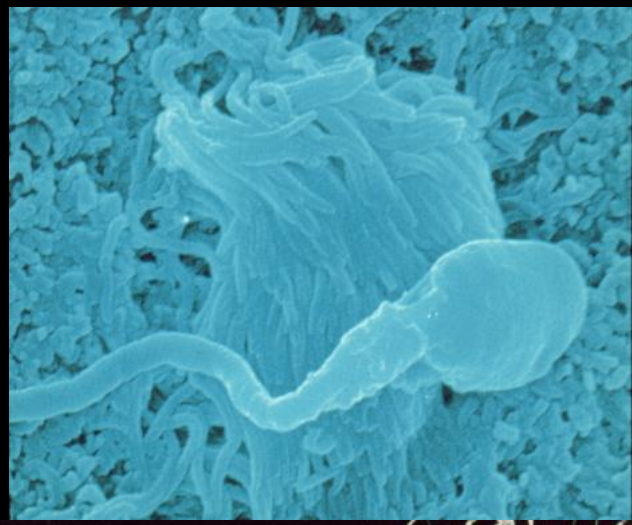


# Sexual Reproduction

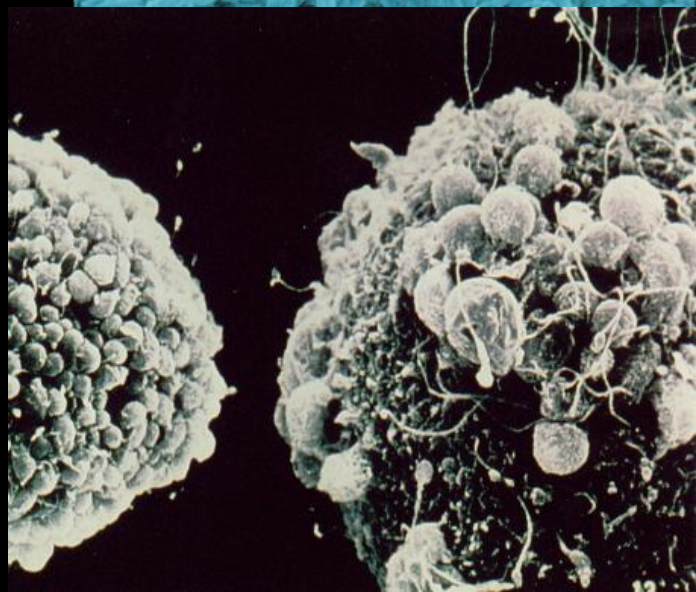
We have similar characteristics to our parents due to genetic information being passed down in genes through gametes:



The human egg and sperm cell ("GAMETES") contain 23 chromosomes each.



When fertilisation happens the gametes fuse together to make a single cell called a ZYGOTE. The zygote has 46 chromosomes (23 pairs) and contains information from each parent.



# Sexual vs. Asexual reproduction

## Sexual reproduction:

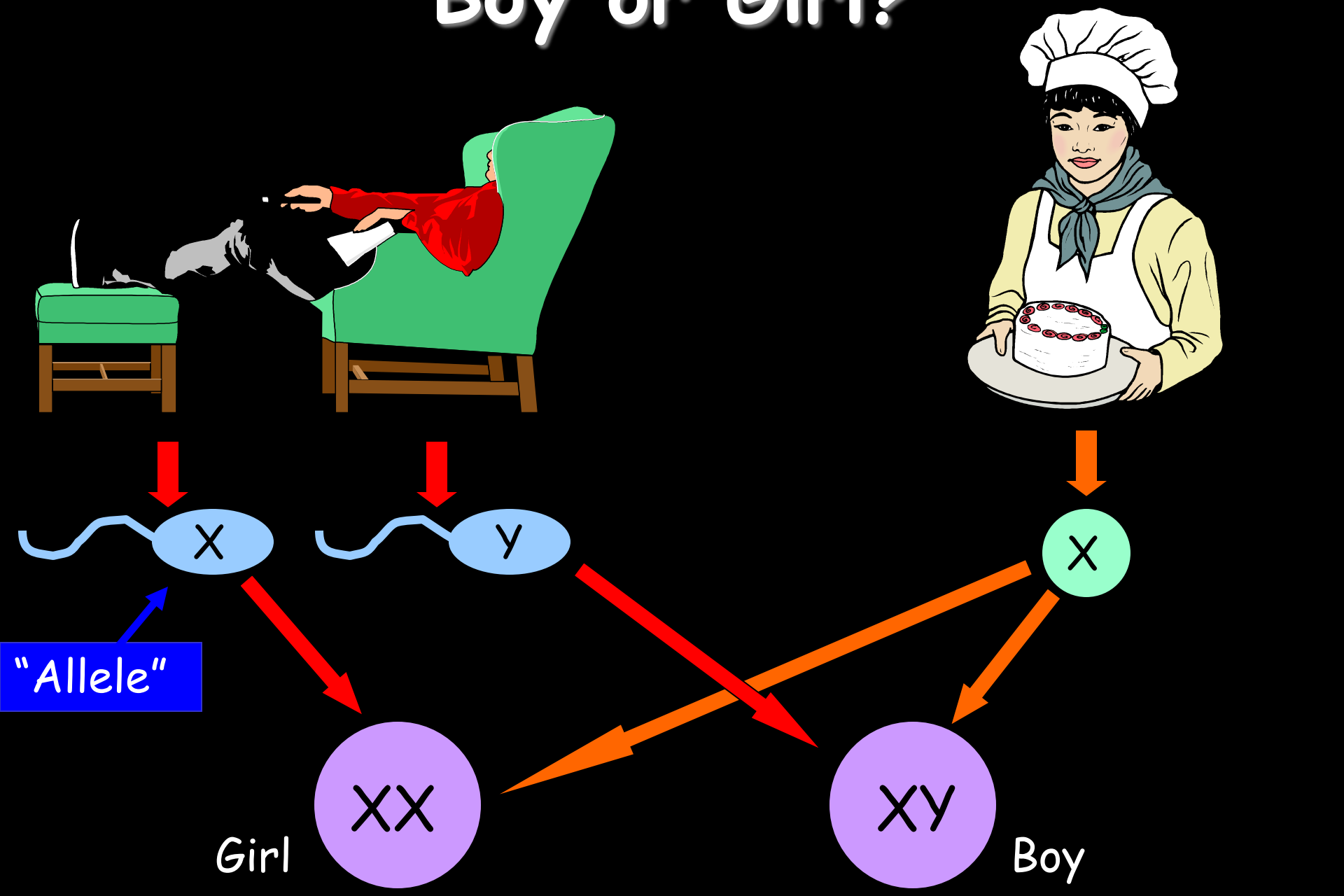
- 2 parents are needed
- Offspring will have "pairs" of chromosomes
- This will cause genetic variation

## Asexual reproduction:

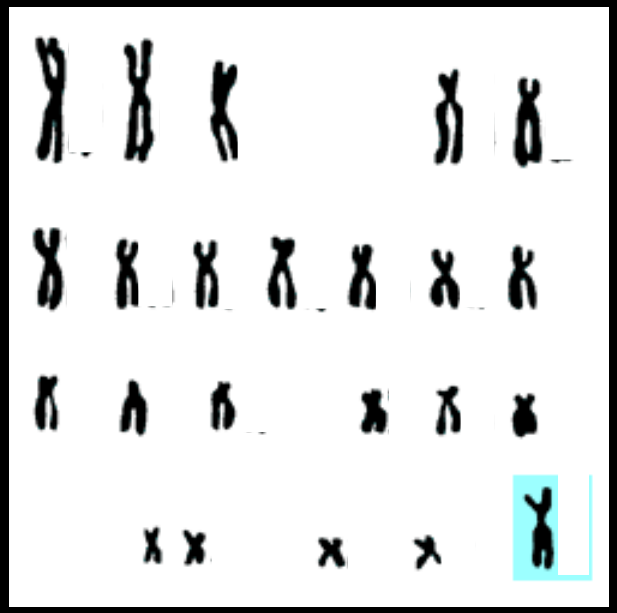
- Only 1 parent needed
- Offspring are GENETICALLY IDENTICAL to parent ("clones")

*"Snuppy" – the first cloned dog (Aug 05)*

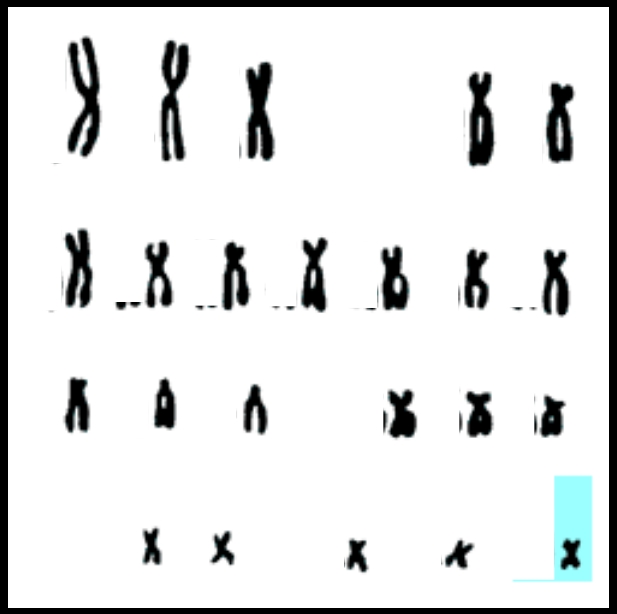
# Boy or Girl?



# Boy or Girl?



Mother



Father

Son

Daughter

# Androgen

Androgen is a hormone that controls the development of male reproductive organs:

***Step 1:*** *An egg is fertilised by an X chromosome and a Y chromosome*

***Step 2:*** *Testes start to develop due to the presence of a Y chromosome*

***Step 3:*** *The testes start producing androgen*

***Step 4:*** *Andogen causes male reproductive organs to grow*

Sometimes the Y chromosome is present but androgen is not detected. This causes the development of female reproductive organs but the individual is genetically a male and unable to reproduce.

# Eye colour

In eye colour the brown eye allele is dominant, so we call it B, and the blue eye is recessive, so we call it b:

BB

*Homozygous brown-eyed parent*

Bb

*Heterozygous brown-eyed parent*

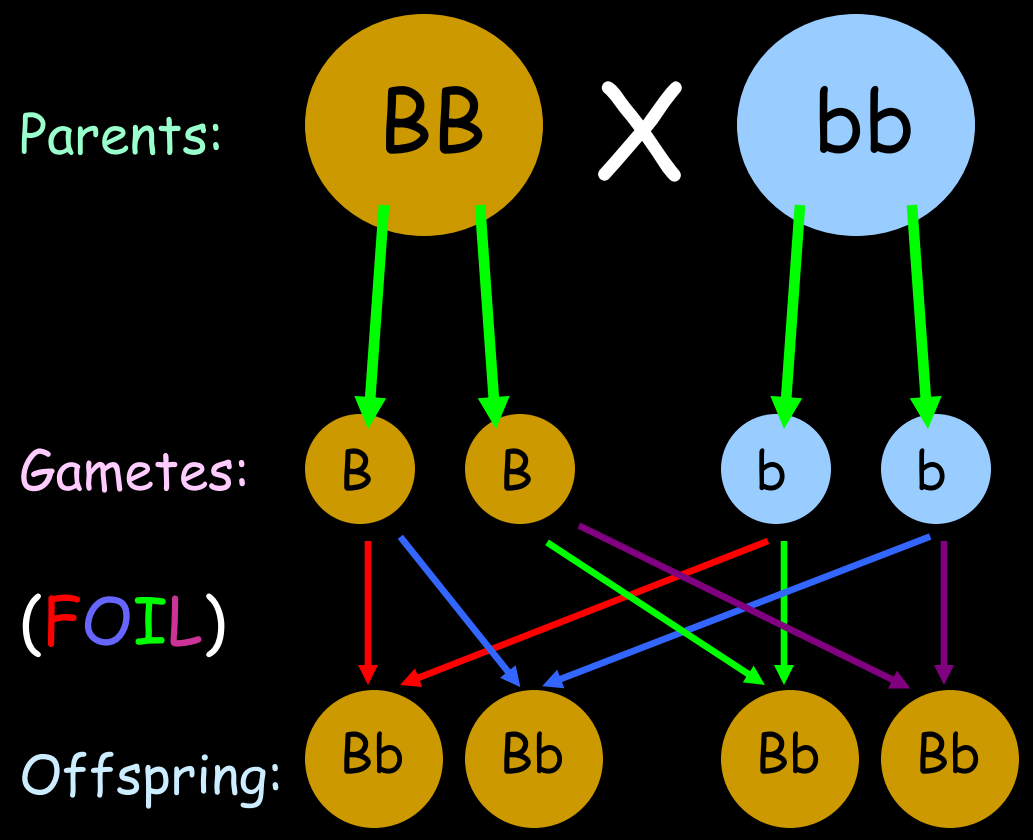
bb

*Blue-eyed parent*

***What would the offspring have?***

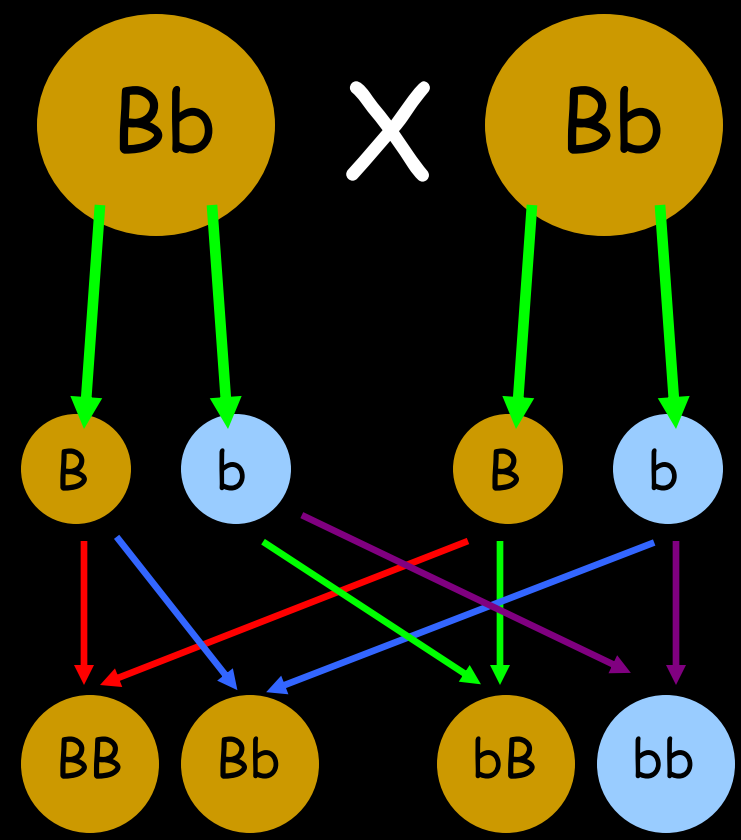
# Eye colour

## Example 1: A homozygous brown-eyed parent and a blue-eyed parent:




*All offspring have brown eyes*

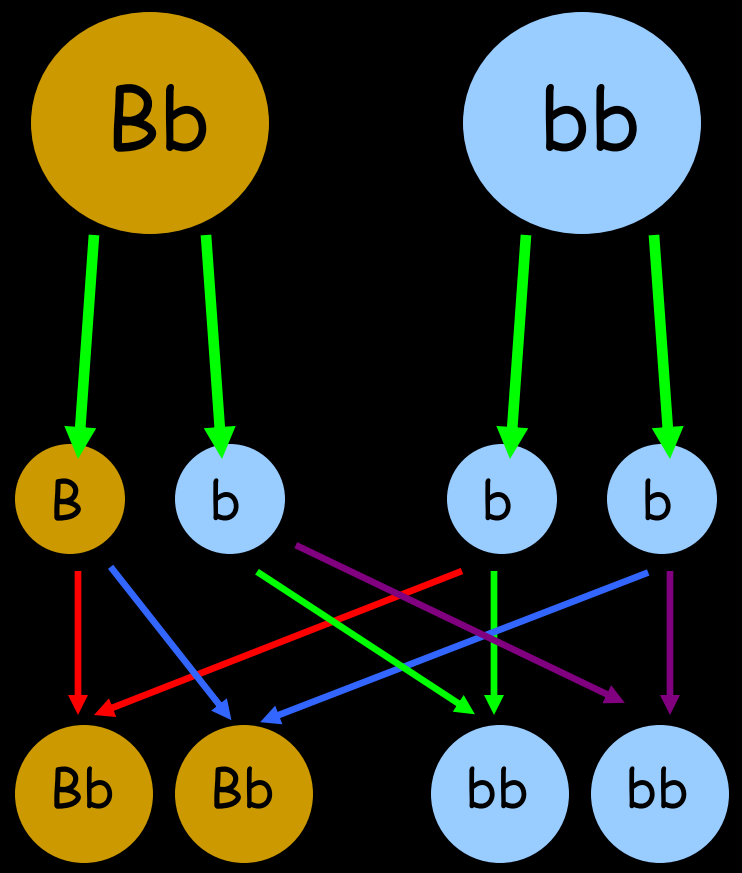
## Example 2: 2 heterozygous brown-eyed parents




*25% chance of blue eyes*

# Eye colour

Example 3: A heterozygous brown-eyed father and a blue-eyed mother:




*Equal (50%) chance of being either brown eyed or blue eyed.*

# Another method

Example 3: A heterozygous brown-eyed father and a blue-eyed mother:

| Mother \ Father | B | b |
|---|---|---|
| b | Bb | bb |
| b | Bb | bb |

# Example questions

1) In mice, white fur is dominant. What type of offspring would you expect from a cross between a heterozygous individual and one with grey fur? Explain your answer with a genetic diagram.

2) A homozygous long-tailed cat is crossed with a homozygous short-tailed cat and produces a litter of 9 long-tailed kittens. Show the probable offspring which would be produced if two of these kittens were mated and describe the characteristics of the offspring (hint: work out the kitten's genotype first).

# Inherited diseases

1) Cystic fibrosis – a disease that causes thick and sticky mucus to coat the lungs, gut and pancreas. It's caused by recessive alleles:

Ff X Ff

2) Huntingdon's disease – a disease of the nervous system that causes shaking and eventually dementia. It's caused by a dominant allele:

Cc X cc

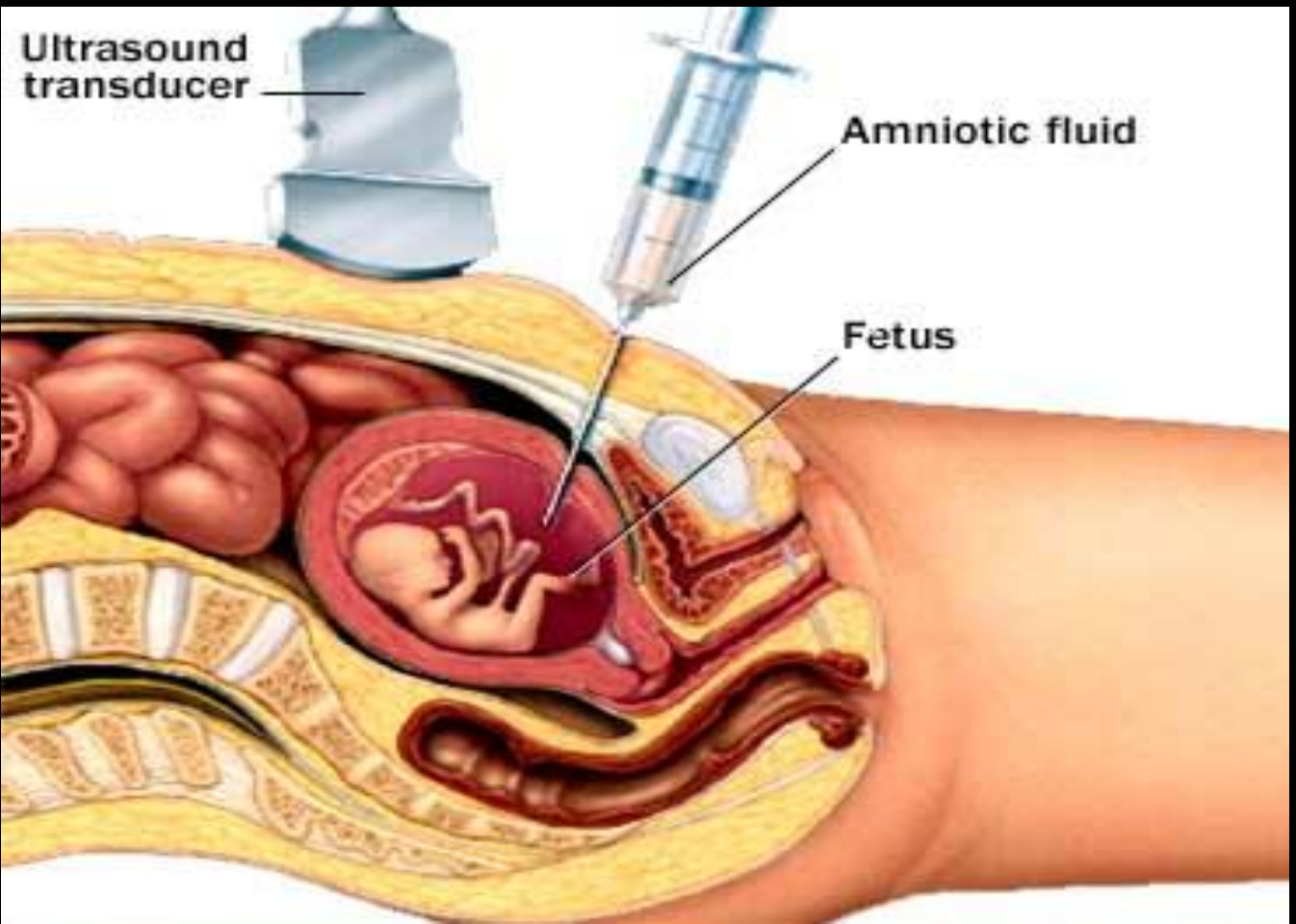
3) Sickle cell anaemia – a disease that alters the shape of red blood cells, thereby reducing their oxygen capacity, causing weakness and anaemia. It's caused by recessive alleles:

Ss X Ss

# Genetic testing

It is now possible to test individuals before they are born for any faulty alleles. There are two main methods:

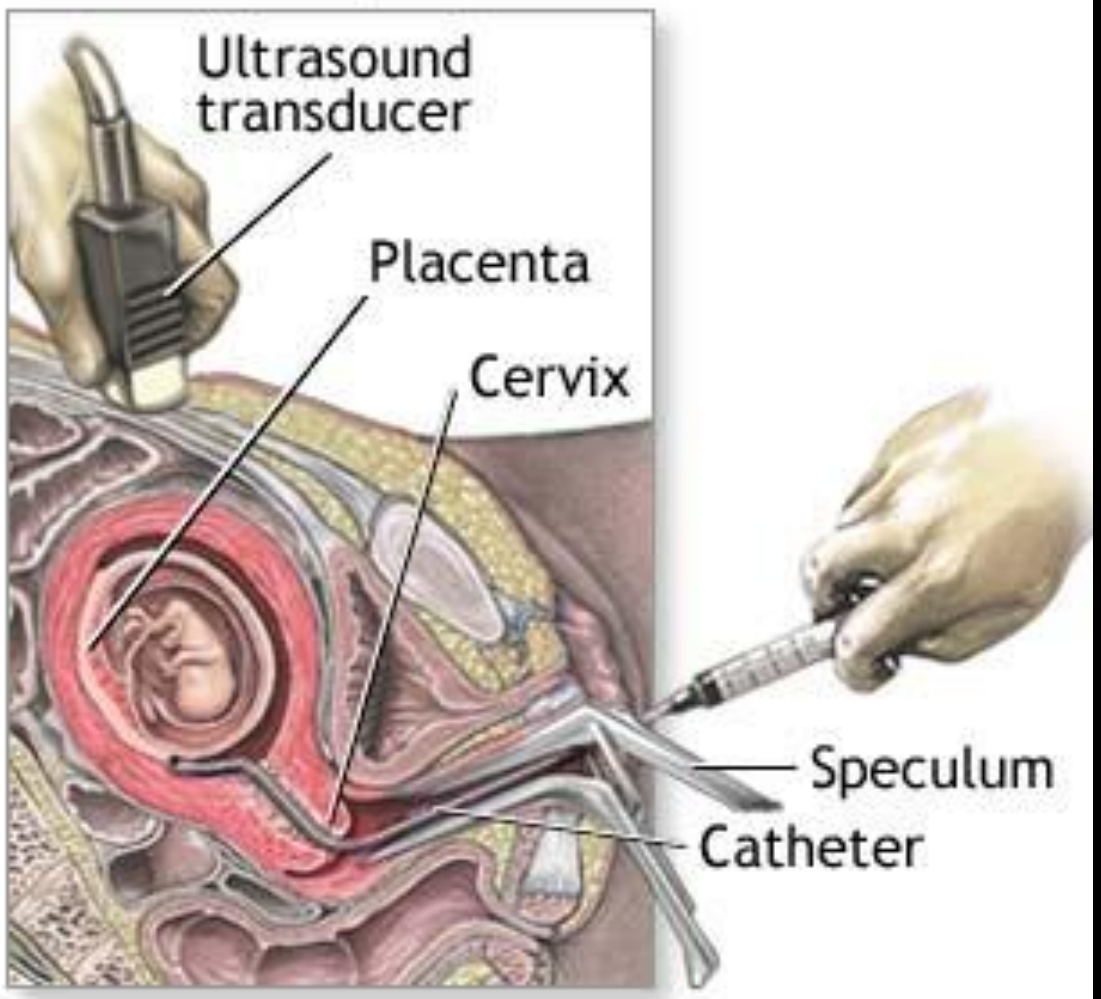
1) Amniocentesis testing:




- Done at 14-16 weeks
- 0.5% chance of miscarriage
- Small chance of infection

# Genetic testing

2) Chorionic villi testing:




- Done at 8-10 weeks
- 2% chance of miscarriage
- Virtually no chance of infection

# Genetic testing

Possible outcomes:

| Outcome | Test result | Reality |
|---|---|---|
| True positive | Fetus has the disorder | Fetus has the disorder |
| True negative | Fetus does not have the disorder | Fetus does not have the disorder |
| False positive | Fetus has the disorder | Fetus **does not** have the disorder |
| False negative | Fetus does not have the disorder | Fetus **has** the disorder |

# The Ethics of Genetic Testing

Genetic testing

- Eliminating genetic diseases
- Aborting deformed children
- Genetic information/ profiles stored on computer
- Predicting future diseases

# Embryo selection

Another way of preventing babies born with genetic disorders is embryo selection.

Basically, embryos are harvested from the mother and fertilised in a lab by the father's sperm (IVF). Healthy embryos are then impmanted back into the mother. This procedure is called pre-implantation genetic diagnosis (PGD).

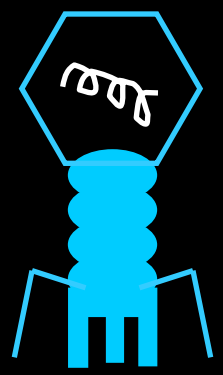
Genetically engineered people in TV:


Mutant X, a team of genetically engineered mutants.

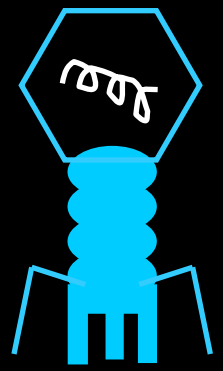
Khan, from Star Trek 2: The Wrath of Khan

# Gene Therapy

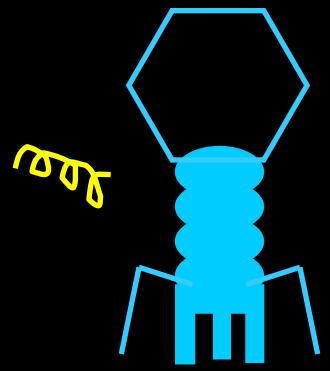
Gene therapy is when diseases are treated by modifying a person's genome. For example, take cystic fibrosis:



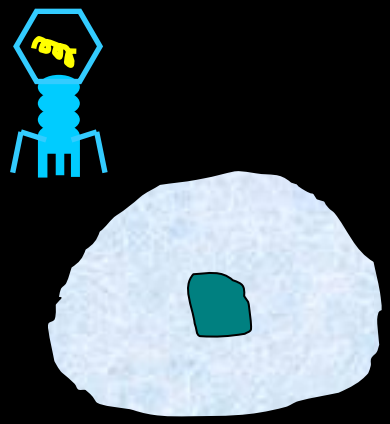
1) Take a disease-causing virus



2) Remove the genetic material that causes excessive mucus to be produced



3) Insert the genetic material that stops cells producing excess mucus



4) The virus then works on the cells in the lung

# Gene Therapy

Gene therapy is when a disease is treated by modifying a person's "genome" (genetic makeup). Consider the example of cystic fibrosis, a disease causing lung problems:

The patient is anaesthetised

↓

They inhale a "vector virus" carrying a normal gene

↓

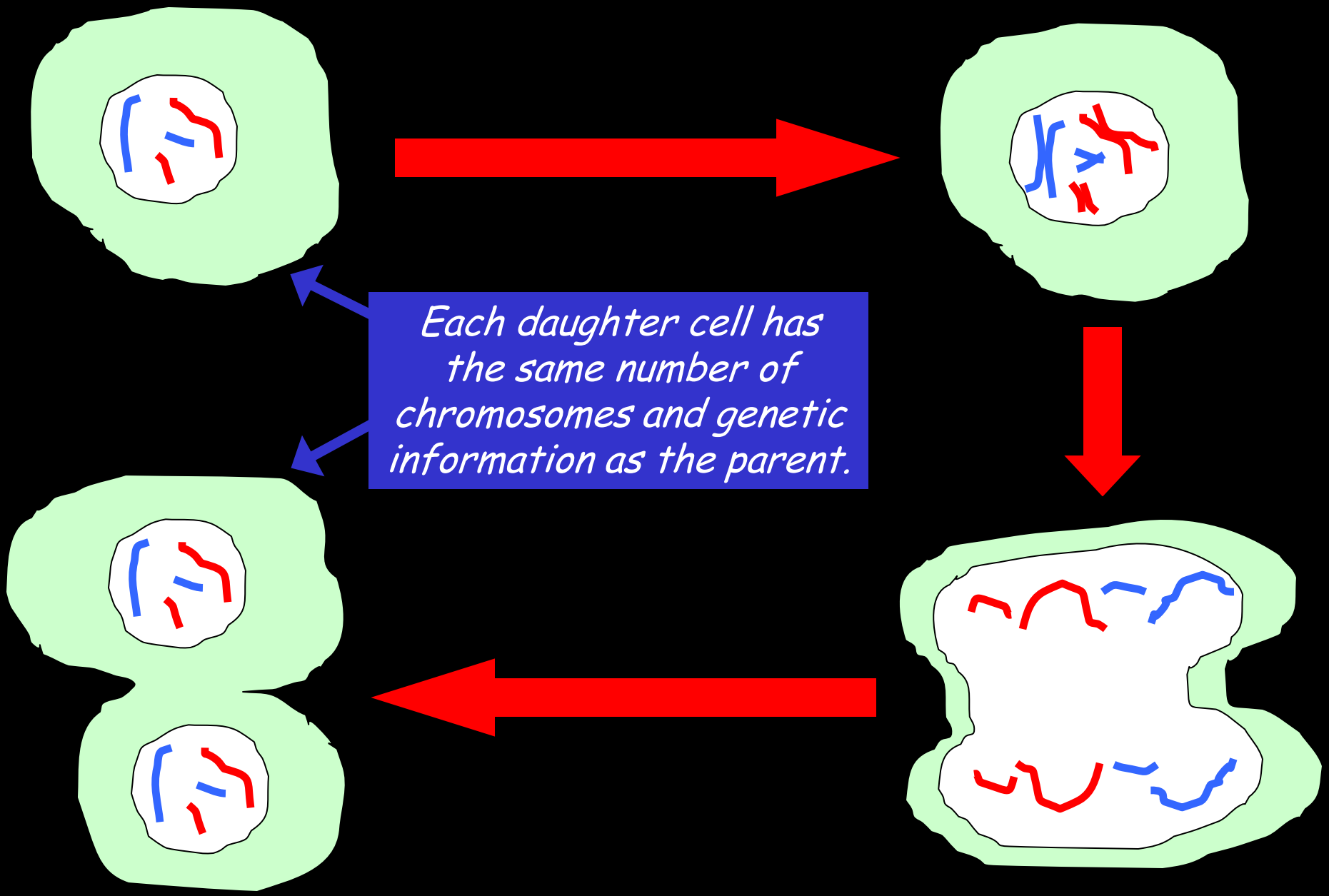
The normal genes enter lung cells

↓

Patients feel better for around 4 weeks

Gene therapy can also be used to treat breast cancer but it is very expensive. What are the ethical issues?

# Asexual reproduction in cells



# Cloning Animals

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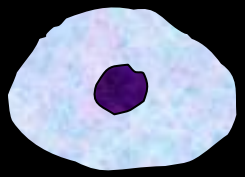
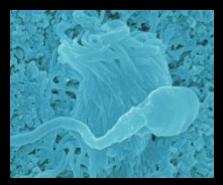
Host mother

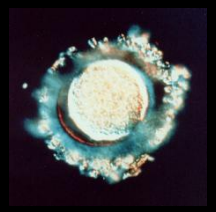
Clone



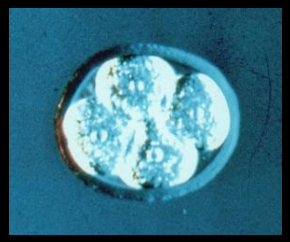
# Stem cell research

Stem cells are cells that have not yet specialised:

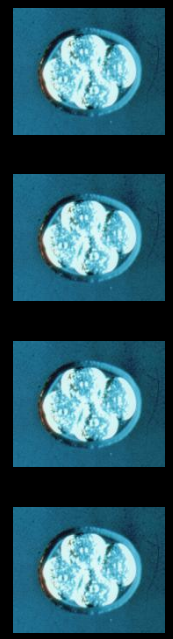




Egg and sperm



Embryo



Cloned embryos

These stem cells have the potential to develop into any kind of cell. The rest of the embryo is destroyed. Most of these embryos come from unused IVF treatments.

**The ethical issue:**

Should these embryos be treated as humans?

# Making decisions

Some questions cannot be answered by science and need to be considered on ethical grounds.

*Factors that might influence a decision:*

- Beliefs/religion
- What does "the right thing" mean?
- "Playing God"
- Risks – acceptable or unacceptable?
- Social and economic contexts