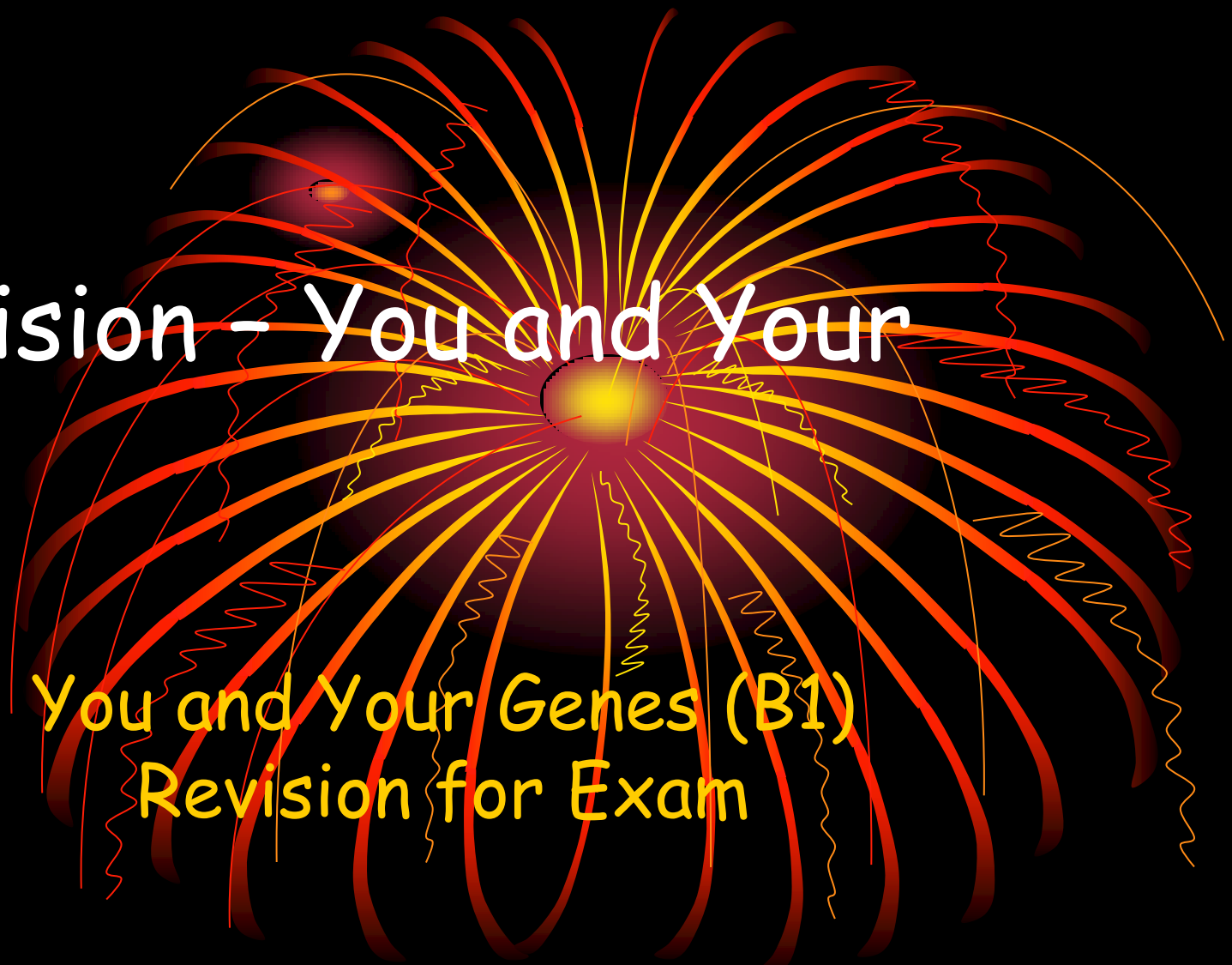
B1 Revision – You and Your Genes



You and Your Genes (B1)
Revision for Exam

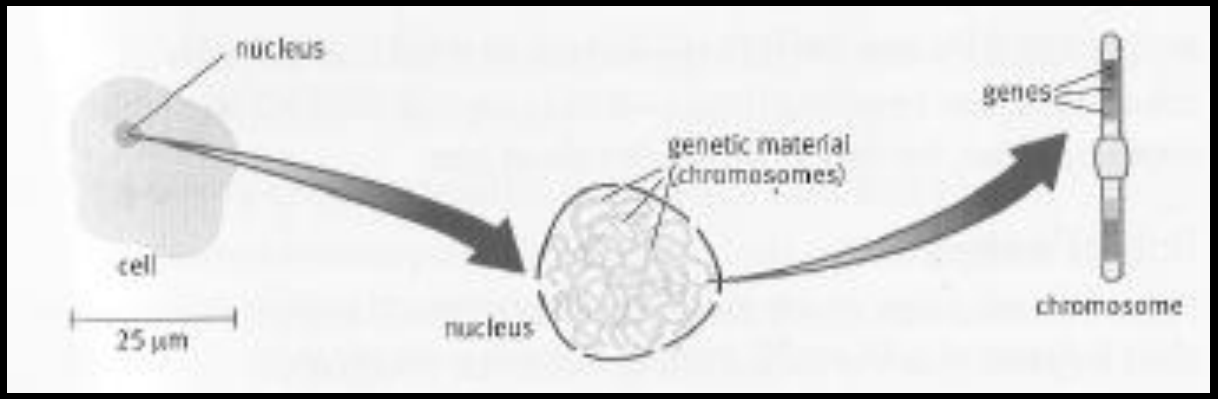
What makes us all different?

Organisms inherit information from their parents. This controls how they develop, so children look a lot like their parents. People share many common features and the differences between people are very small. These differences make us unique.

Most features are also affected by the environment e.g. Weight, height, skin colour

Chromosomes are made of DNA molecules

All living things are made of cells

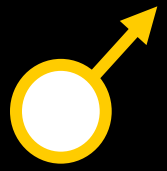


Different proteins in the body do different jobs. **H:** They can be structural (to build the body) or enzymes (to speed up chemical reactions in the body)

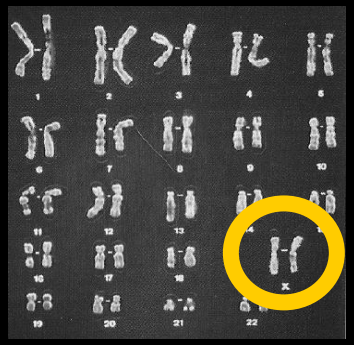
Genes are instructions which tell the cell how to make proteins

What makes a person male or female?

A fertilised human egg cell contains 23 pairs of chromosomes. Pair 23 are the sex chromosomes.

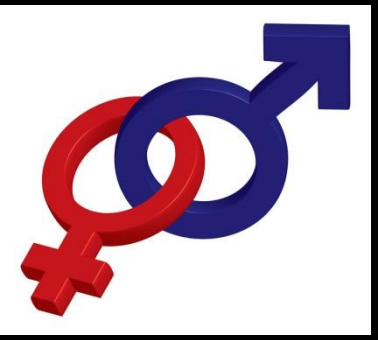


Males have an X and Y chromosome - **XY**



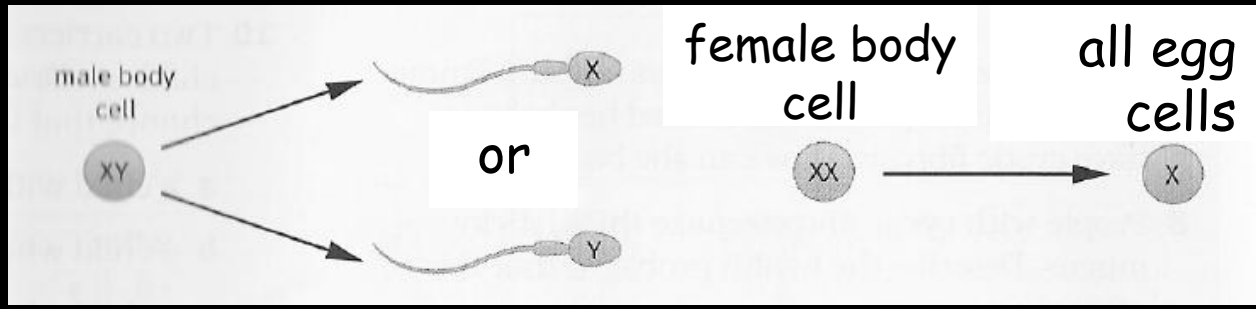
Females have two X chromosomes - **XX**

H: The SRY gene on the Y chromosome causes the testes to produce the male sex hormone, **androgen**, which makes the embryo develop into a male.



male body cell

sperm



50% chance that a baby will be a boy or a girl

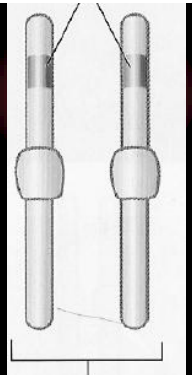
50% chance that an X or Y chromosome fertilises an egg

Inheritance and Alleles

Parents pass on genes in their **sex cells**, which contain copies of **half** the parent's chromosomes.

Chromosomes come in pairs. They carry the same genes in the same place on each chromosome, so genes also come in pairs

same gene



chromosome pair

Alleles
= different versions of genes in a pair

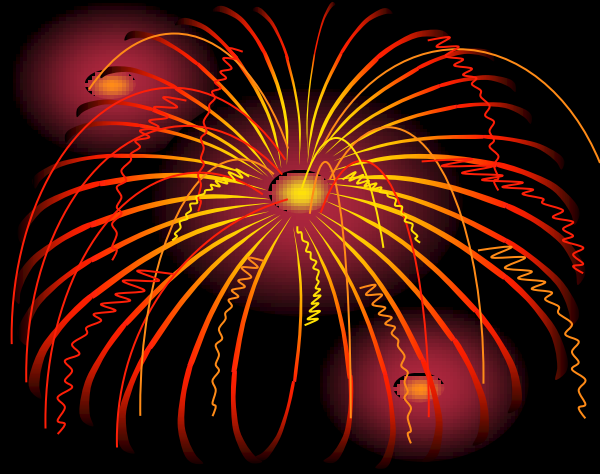
Dominant
-in charge
-only need 1 copy to have its feature

Recessive
- must have 2 copies of this allele to have its feature

☺ sex cells get one chromosome from each pair the parent has
☺ cannot predict which egg & sperm cells will meet

Example:
T = dominant allele for straight thumbs
t = recessive allele for curved thumbs
TT = straight thumbs Tt = straight thumbs tt = curved thumbs

Huntington's Disorder



Symptoms (Affects the nervous system, later in life e.g. 40-50yrs old)

- -forgetful → difficulty understanding things
- -twitching of muscles → unable to control movements
- -eventually a fatal condition

An inherited condition, caused by a **dominant allele**

Worked Example:

H = dominant allele for Huntington's disorder

h = recessive allele (non-Huntington's)

If the man is Hh, he will get the disorder. If the woman is hh, what percentage of their children will inherit the condition?

father **Hh**

H h

Mother **hh**

	h	h
h	Hh	Hh
h	Hh	hh

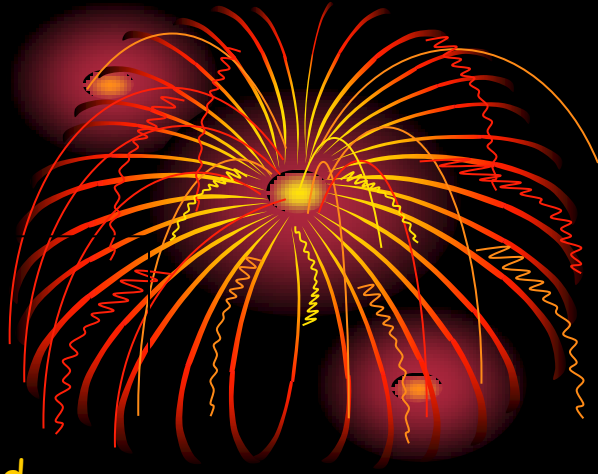
Hh affected, so,

50% will inherit Huntington's

Cystic Fibrosis

CF is an inherited illness
- -affects breathing & digestion
- -cells that make mucus produce mucus which is too thick, so it blocks the lungs & tubes that take enzymes from the pancreas to the gut

- breathlessness
- chest infections
- food not digested properly
- short of nutrients



If both parents are carriers there is a 1 in 4 chance their child will have CF

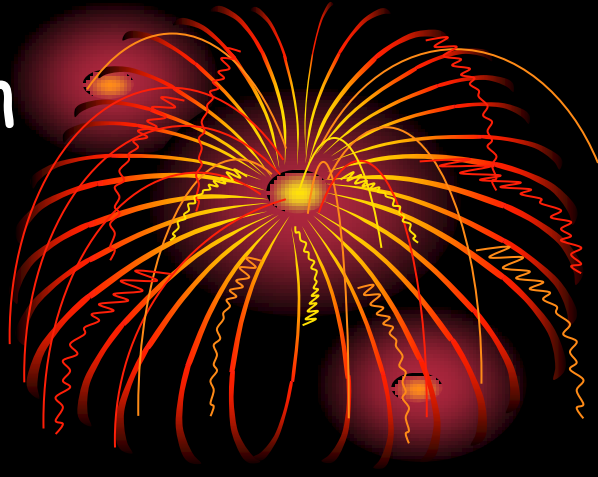
Caused by a faulty **recessive allele** (f), so a person may be a **carrier** of the faulty gene (Ff), but not have cystic fibrosis

1 in 25 people in the UK carries the faulty recessive allele. Would you want to know if you're a carrier?

father (carrier) Ff

mother (carrier) Ff	F	f
F	FF	Ff
f	Ff	ff

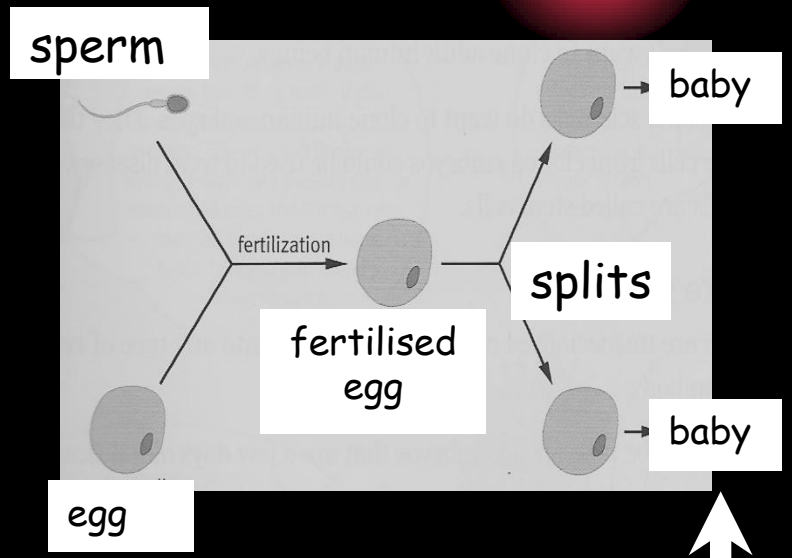
Asexual and Sexual Reproduction



Asexual reproduction is performed by only one parent & the offspring are genetically identical to the parent. They are **clones**.

Any variation between them is caused by differences in their environment

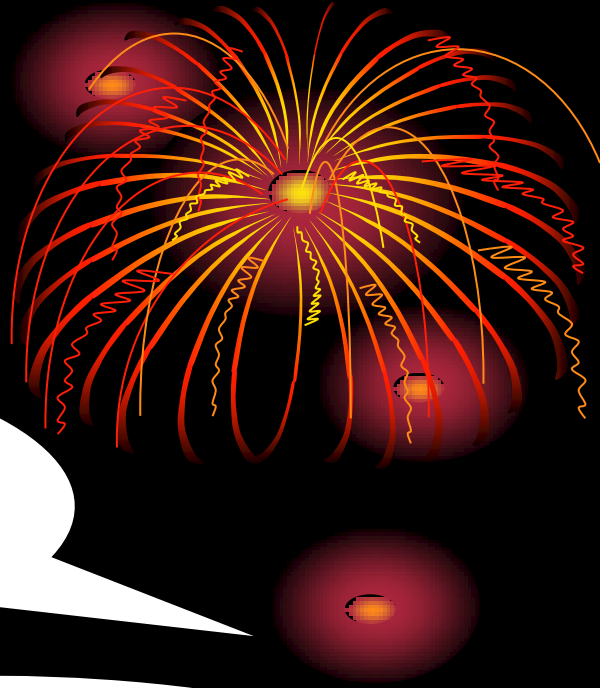
Plants keep some **unspecialized cells** all their lives, which can become anything that the plant might need. They can be used for asexual reproduction.



Most animals use sexual reproduction, requiring 2 parents & producing offspring which contain a random mix of genes from both parents. **Clones** are sometimes produced when identical twins are formed. These are natural clones. Artificial clones can be made (Dolly)

Genetic Testing of Adults

Genetic testing is when an individual is tested for the presence of a particular allele that may cause a genetic disorder.



Who would want to be tested?

Someone with a family history for a serious genetic disorder.

Like Huntington's disorder or cystic fibrosis?

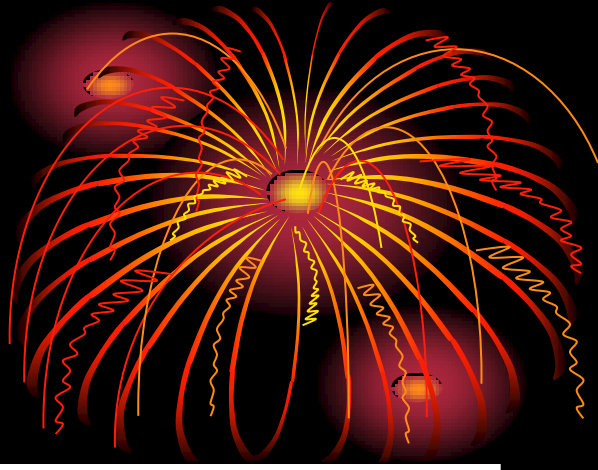
Yes. It helps them to plan their lives. Such as whether to have children of their own or not.

Are there any disadvantages?

Yes. Testing can be stressful and in some cases, a positive result may make it difficult to get a mortgage, job or life insurance.

Genetic testing of foetuses

A couple may decide to have a genetic test on the foetus during pregnancy, if they know there is a risk they may pass on a serious genetic disorder to their children.



How is this done?

Doctors collect cells from the foetus during pregnancy, using an amniocentesis test, and then examine the genes of the foetus.

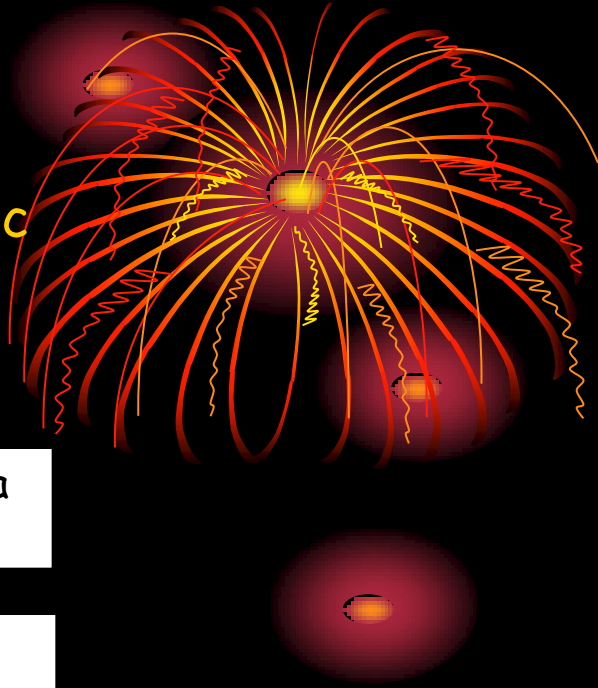
What happens next?

If the child will inherit the genetic disorder, the parents may choose to have a **termination** (abortion).

Are there any risks from testing?

Yes. There is a risk of miscarriage during the amniocentesis test and the results are not 100% reliable.

Genetic testing of embryos



A couple may decide to use pre-implantation genetic diagnosis (PGD), to avoid passing on a serious genetic disorder to their children, without the possible need for a termination.

The woman is given fertility drugs, so she produces several ova at once.

↓

The ova are collected and mixed with sperm in a dish. Fertilization occurs. This is ***in vitro* fertilisation** (IVF).

↓

The fertilised eggs start to develop into embryos.

↓

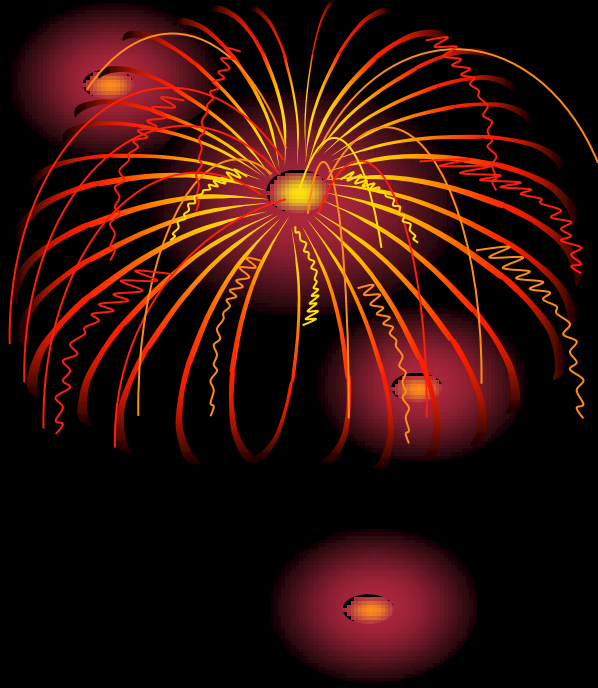
Cells from the embryos are tested for the presence of faulty alleles that cause the genetic disorder.

↓

Only embryos without the faulty alleles are implanted into the uterus of the woman to develop into a baby.

PGD can only be used for families with certain inherited conditions. The Human Fertilisation and Embryology Authority (HFEA) is a committee that decides when PGD can be used.

Genetic Screening



Genetic screening is when a whole population is tested for a particular allele.

The NHS and Government decide whether to use genetic screening based upon:

- the relative costs of screening compared to treatment
- the possible benefits to individuals and the population

Who should have access to the information?

Individuals
Make informed decisions.
May not want to know.
Right to choose.

Insurance Companies
Assess if person is a higher risk.
Charge higher premiums or refuse insurance.

Employers
Assess if employee will be fit to work in the future.
May lead to discrimination of individuals.

Gene Therapy

Gene therapy involves introducing normal functional alleles into the cells of patients with a genetic disorder. In the future, it is hoped gene therapy will cure genetic disorders such as cystic fibrosis.

Find the gene that causes the disorder on a chromosome.

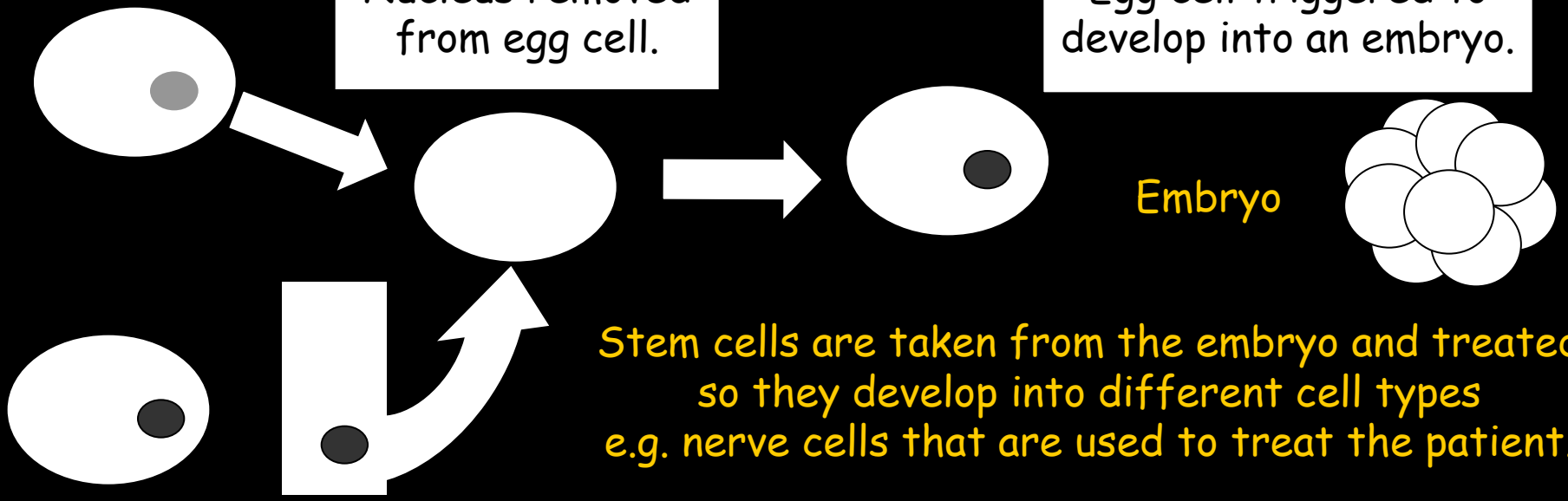
Cut out the normal allele from another chromosome and make lots of copies of it.

Introduce the normal alleles into cells of the patient.

Therapeutic Cloning

Stem cells are unspecialised cells that can develop into different cell types. Scientists believe stem cells could be used to treat certain degenerative diseases e.g. Parkinson's.

Embryonic stem cells are most useful because they can develop into any cell type found in the body.



Stem cells are taken from the embryo and treated so they develop into different cell types e.g. nerve cells that are used to treat the patient.

These cells have the same genes as the patient, so would not be 'rejected'.