Inheritance, cystic fibrosis and genetic screening

Genetic terms, monohybrid and sex-linked inheritance, pedigree diagrams, cystic fibrosis, and genetic screening with its ethical issues.

Genetic terms

Term	Meaning
Gene	A sequence of DNA bases that codes for a polypeptide
Allele	A version of a gene
Genotype	The alleles an organism has
Phenotype	The observable characteristics, resulting from genotype and environment
Dominant	An allele that is expressed in the **heterozygote**
Recessive	An allele that is expressed only in the **homozygote**
Codominant	Both alleles are expressed in the heterozygote (e.g. human ABO blood groups, A and B)
Homozygous	Two identical alleles
Heterozygous	Two different alleles

Monohybrid inheritance

A **monohybrid cross** follows one gene with two alleles. Use a **Punnett square**.

> **TRY IT**
>
> Two heterozygous parents ($Aa \times Aa$) have children. Give the genotype and phenotype ratios, with a recessive.

Genotypes: 1 AA : 2 Aa : 1 aa. Phenotypes: **3 dominant : 1 recessive**.

A **test cross** crosses an individual showing the dominant phenotype with a homozygous recessive (aa). If any offspring show the recessive phenotype, the parent was heterozygous.

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Sex linkage

The **X chromosome** carries many genes that are not on the **Y** chromosome. Males (XY) have only one allele, so a **recessive** allele on the X is expressed in males. Females (XX) need two copies to be affected, but can be **carriers**.

Red–green colour blindness is a sex-linked recessive condition (X^b). A colour-blind son has a carrier or affected mother; he cannot inherit it from his father (his X comes from his mother). A carrier mother (X^BX^b) and normal father (X^BY) have sons with a 50% chance of colour blindness, and none of the daughters are affected (50% are carriers).

Pedigree diagrams

Squares are males and circles are females. Shaded symbols show the affected.

- If two **unaffected** parents have an **affected** child, the allele is **recessive**, and both parents are carriers.
- If an **affected** child has an affected parent, and the trait appears in every generation, it may be dominant.
- If many more males than females are affected, and the trait passes through carrier females, it is **sex linked**.

> **NOTE**
>
> Give the genotypes in a pedigree, and give the probability, using the multiplication rule: the chance of two independent events is the product of the chances.

Cystic fibrosis

Cystic fibrosis (CF) is caused by a **recessive allele** of the **CFTR gene** (on chromosome 7), most often a **deletion of three bases** (ΔF508). It codes for a **chloride ion channel** in epithelial cell membranes. The faulty protein does not reach the membrane or does not work.

Without the channel, chloride ions do not leave the cells, so **less water leaves by osmosis**, and the **mucus becomes thick and sticky**.

- **Gas exchange system**: mucus blocks the airways and traps bacteria, leading to infection; the alveoli are damaged.
- **Digestive system**: mucus blocks the **pancreatic duct**, so digestive enzymes do not reach the small intestine; absorption of nutrients is poor.
- **Reproductive system**: the tubes (the vas deferens in men) can be blocked, so men are often infertile; mucus in the cervix in women reduces fertility.

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Genetic screening

Genetic screening tests people for a particular allele.

- **Carrier testing** identifies healthy people who carry a recessive allele (e.g. for CF), so couples can make an informed choice.
- **Pre-implantation genetic diagnosis (PGD)**: embryos made by **in vitro fertilisation** are tested at the 8-cell stage by removing one cell, and only unaffected embryos are implanted.
- **Prenatal testing**: **amniocentesis** (a sample of amniotic fluid containing fetal cells, from about 15 weeks, with about a 1% miscarriage risk) and **chorionic villus sampling** (a sample of tissue from the placenta, from about 11 weeks, a slightly higher risk). The DNA is analysed.

Implications and ethical issues

- Benefits: informed decisions, early treatment, and prevention of suffering.
- Issues: the risk of miscarriage from invasive tests, the **psychological stress** of the result, decisions about **abortion** or discarding embryos, **"designer babies"**, **discrimination** by employers or insurers, and **privacy** of genetic data. A false result is possible.
- Different **viewpoints**: **religious** views may hold that life begins at conception, so selection is wrong; **moral** views weigh the suffering of the child against the rights of the embryo; **social** views consider the cost to society and the stigma on those with the condition.

> **When asked to discuss an ethical issue, give more than one viewpoint and say who is affected. Do not just give your own opinion.**

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