BioMedical Admissions Test (BMAT)

Section 2: Biology

Topic B4 - Inheritance

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Genetic Terms

Gene	Short sequences of DNA on chromosomes which code for a protein
Allele	Different versions of genes. For example, the gene for blue eyes and the gene for brown eyes are 2 different alleles of the same gene.
Dominant	Only one dominant allele is needed to determine the phenotype
Recessive	Only determines the phenotype if there are two copies of this allele
Heterozygous	If an individual has two different alleles of that gene, e.g. Tt
Homozygous	If an individual has two alleles of a gene that are the same, e.g. TT
Genotype	The combination of alleles e.g. Tt
Phenotype	The visible characteristics as a result of the alleles
Chromosomes	Condensed strands of DNA. All our genetic material is held in 23 pairs of chromosomes

Genetic Diagrams

You need to understand how to **create and analyse monohybrid crosses** (i.e. for a single gene)

Genetic diagrams can help you determine:

- The likelihood of offspring being male or female.
- The probability of offspring having a certain characteristic.

> ***Exam Tip*** - Simple genetic diagrams help you determine the likelihood of offspring having a certain characteristic, when a **single gene** controls that characteristic.
>
> However, it is often more complicated than that; **one characteristic can be influenced by several different genes**. It is likely that the BMAT exam will only give you genetic diagrams with characteristics controlled by a single gene, but keep this point in mind as it could be useful in identifying trick answers.

Monohybrid Crosses

A **punnett square** based on a monohybrid cross is a diagram that shows you the possible genetic combinations from two parents for a single trait for simplicity. However, it is important to note that most phenotypic features are a result of the interaction of many genes, rather than a single gene inheritance.

When drawing punnett squares, letters are often used to represent alleles.

- An uppercase letter is used to represent the **dominant** allele (e.g. **A**)
- A lowercase letter for the **recessive** allele (e.g. **a**).

In order to display a recessive characteristic, the organism must be **homozygous recessive** for that trait. Both **homozygous dominant** and **heterozygous** individuals will display the dominant characteristic.

Example

Let's look at this using the example of the gene for eye colour.

- ➔ The father is **homozygous recessive** and has **blue** eyes. This means his genotype is **aa**.
- ➔ The mother is **heterozygous** and has **brown** eyes. This means her genotype is **Aa**.

To make a punnett square, all you need to do is draw a grid like the one below, filling in the mother's and father's genotypes as given, and then combining the alleles to complete the squares for the offspring.

		Father's genotype	
		a	a
Mother's genotype	A	Aa	Aa
	a	aa	aa

Interpretation

We can use the punnett square above to calculate the probability of a child having a certain phenotype or genotype.

- Of the 4 possible combinations, 50% are homozygous recessive and 50% are heterozygous. This means that **at every pregnancy,** there is a **50% probability** of the child having **'Aa' alleles**, and a **50% probability** of the child having **'aa' alleles.**
- Since the homozygous recessive genotype ('aa') causes individuals to have blue eyes, there is a **50% probability** that the offspring will have that phenotype. Similarly, given that the heterozygous genotype ('Aa') results in the brown eyes, there is a **50% probability** that the offspring will have **brown eyes.**

We can also create a punnett square for the X and Y chromosomes.

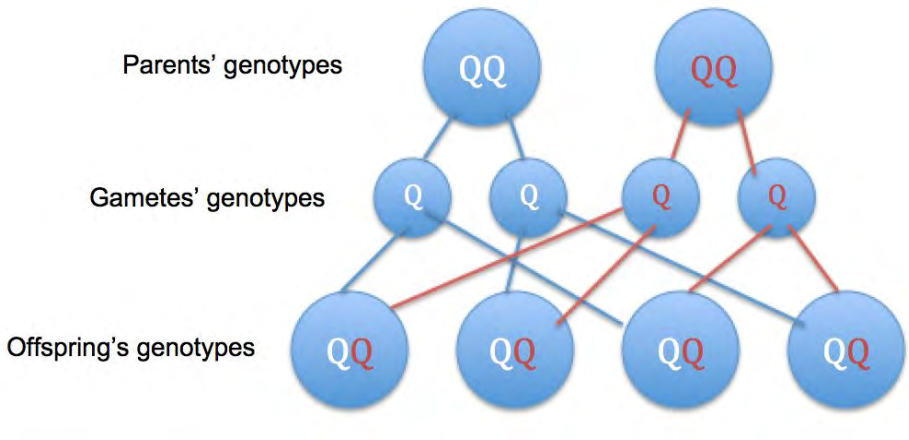
		Father's genotype	
		X	Y
Mother's genotype	X	XX	XY
	X	XX	XY

From this diagram, we can see that there is **always a 50% probability that the parents' offspring will be male** (with the XY phenotype) **or female** (with the XX phenotype).

Other genetic diagrams

A **genetic diagram** is an umbrella term. As well as a punnett square, we can also determine monohybrid crosses using family trees.

Assuming that the alleles for a certain characteristic are denoted by q, and Q represents the dominant allele, this diagram represents a situation where both parents are **homozygous dominant**. If both parents are homozygous dominant for a trait, the offspring will **always be homozygous dominant** and express the same trait.



It's worth noting here that the same is true for 2 parents **homozygous recessive** for a particular trait: all offspring will also be homozygous recessive for the trait. The absence of the dominant allele means that the only possible combination for offspring is 2 recessive alleles.

Inherited conditions

Polydactyly

- Caused by a **dominant** allele, meaning that inheriting **one** disease allele is enough to cause the condition.
- Polydactyly is a condition where babies are born with **extra fingers and toes**.
- If a child has the disease allele, at least one parent must carry the allele. Since it is a dominant alle, it therefore follows that this parent must also have the disease.
- This punnett square outlines how the dominant allele for polydactyly (D) is inherited:

		Father's genotype	
		d	d
Mother's genotype (has polydactyly)	D	Dd	Dd
	d	dd	dd

- You can see from the diagram that 50% of the offspring will be heterozygous, and 50% will be homozygous dominant. This means there is a **1:1 ratio** of offspring without polydactyly to offspring with polydactyly, when only one parent has the disease.

Exam Tip - Make sure you understand **ratios** relating to genetic diagrams. For example, if each box in a punnett square is one child:

- If 3 children out of the 4 (75%) inherit the condition,there is a 3:1 ratio of offspring with the condition to offspring without the condition.
- If 1 out of the 4 (25%) inherit the condition, there is a 1:3 offspring of ratio with the condition to offspring without the condition.
- As in the example for polydactyly, if 2 out of the 4 offspring (50%) inherit the condition, then there is a 1:1 ratio of offspring with the condition to offspring without the condition.

Cystic Fibrosis

- Caused by a **recessive** allele.
- Cystic fibrosis involves problems such as difficulty breathing and digestive problems. These problems are due to a **thick sticky mucus** that sufferers produce; it blocks passageways around the lungs and pancreas.
- Unlike a genetic disorder caused by a dominant allele, to acquire cystic fibrosis, **2 copies of the recessive allele** must be inherited. In other words, a sufferer would be homozygous recessive for the disease allele. Therefore, both of the child's parents must

possess at least one of the **recessive alleles**.

- It is possible that both parents are **asymptomatic** if they are both **heterozygous** for the trait. Carrying the recessive allele for cystic fibrosis is not as rare as you might think: 1 in 25 people carry it.

> ***Exam Tip*** - A **carrier** is someone who is **heterozygous** for a trait: they **carry** one recessive allele, and one dominant allele.
>
> For a genetic disorder like cystic fibrosis which is **caused by a recessive allele**, a carrier will not experience symptoms. He or she is **asymptomatic.** If the disorder is caused by a **dominant allele**, however, the individual will show the symptoms of the disorder.

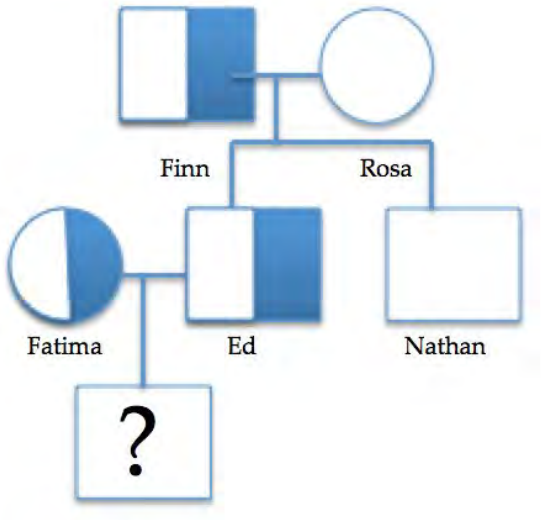
Family Pedigree Diagrams

The inheritance of genetic disorders can be illustrated in a **family tree**.

- **Shaded** squares or circles represent someone who **expresses** the trait.
- **Half-shaded** squares or circles symbolize someone who is a **carrier** (heterozygous)
- Women are always represented as circles, whereas men are always squares.

Worked example

In the diagram below, Finn is a carrier of cystic fibrosis, and so is his son Ed, who also inherits the recessive allele. Many years later, Ed decides to have a child with his partner, Fatima. Since she is also a carrier, what is the probability that their child will have cystic fibrosis? It might help you to draw a punnett square to see this.



Answer: 25%.

Look at the punnett square below for guidance. Let's assume F is the dominant allele, and f is the recessive allele. If both parents are carriers of the trait (heterozygous), you can see from the punnett square that:

- 25% of the offspring will be homozygous dominant.
- 50% will be carriers (heterozygous), like their parents.
- 25% will be homozygous recessive and will therefore have the symptoms of cystic fibrosis.

		Ed's genotype	
		F	f
Fatima's genotype	F	FF	Ff
	f	Ff	ff