

X and Y chromosomes

Sex determination by x and y chromosomes

- ▶ The human body has 23 pairs of chromosomes in its nucleus. 22 of these pairs, called autosomes, control our characteristics. The 23rd pair, known as the sex chromosomes, determine the sex of an individual.
 - In females, the sex chromosomes are the same – **XX**
 - In males, the sex chromosomes are different – **XY**
- ▶ Each parent donates 50% of the offspring's alleles. The female always contributes an X chromosome, whereas the male has a 50/50 chance of contributing either an X or a Y chromosome.

Half of all sperm cells carry an X chromosome, and the other half carry a Y chromosome. In contrast, all egg cells carry an X chromosome.

Some important key terminologies related to genetic inheritance

1. **DNA** is a molecule found in the nucleus of cells that holds the genetic information of an organism.
2. **Chromosomes** are structures that are made of DNA. In meiosis, chromosomes duplicate then get divided up into gametes.
3. **Gametes** are sex cells (in animals they are called egg cells and sperm, in plants they are called egg cells and pollen). They fuse together in fertilisation as part of sexual reproduction.
4. **Genes** are lengths of DNA on chromosomes that code for specific proteins.
5. **Alleles** are different versions of the same gene. Individuals will inherit an allele for each gene from their mother and their father.
6. **Alleles** are **dominant** if they are always expressed, even if there is only one copy. They are written using capital letters.
7. Alleles are **recessive** if they are only expressed when there is two copies (no dominant alleles). They are written using lowercase letters.
8. **Homozygous** individuals have 22 alleles that are the same, e.g. BB or bb.
9. **Heterozygous** individuals have 22 different alleles, e.g. Bb.
10. The **genotype** is the alleles that an organism has for a specific characteristic.

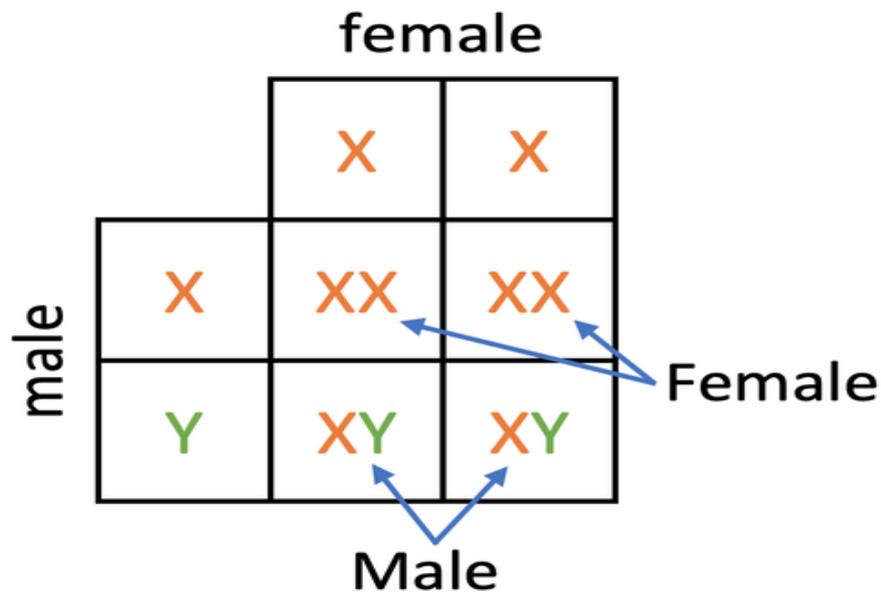
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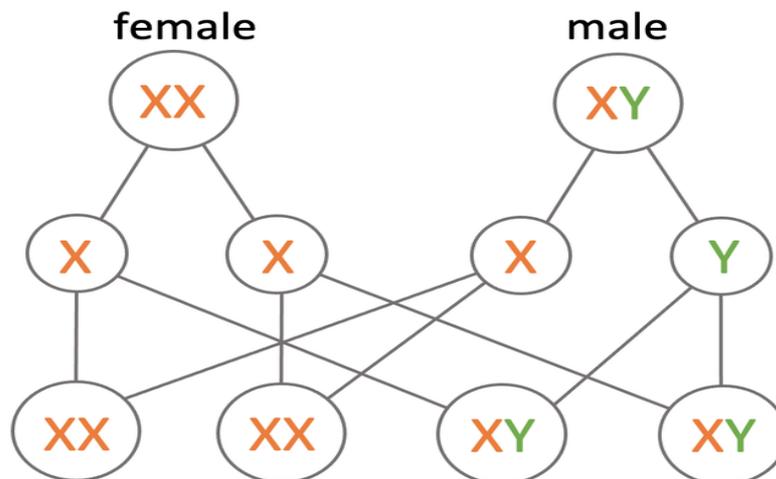
- The **phenotype** of an organism is its observable characteristics that are determined by its genotype.

Genetic diagram or punnet square

- Genetic diagram or Punnett square is used for **determining the sex of offspring**. The male sex cells are sperm and the female sex cells are eggs. Sperm and eggs are both gametes and haploid cells; they have 23 chromosomes.
- A fertilised egg contains 23 pairs of chromosomes or 46 chromosomes in total. Gametes are created by meiosis. Females are XX, which means that they can only produce eggs with an X chromosome in them. Males are XY, which means that half of the sperm will have an X chromosome and the other half will have a Y chromosome.
- The male and female gametes are shown in the Punnett square below.



There is a second diagram that we can draw to show how the sex of offspring is determined. The diagram is shown below, and it works by following the lines.



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- ▶ The top circles in the above diagram show the chromosomes for the female (XX) and the male (XY) that are having the offspring.
- ▶ The next 4 circles below show the gametes that are produced. The female produces two eggs that are both X. And, the male produces two sperm where one is X and the other is Y.
- ▶ These gametes are produced by meiosis. The circles at the bottom show the different combinations for the 23rd pair of chromosomes for the offspring.
- ▶ From the 4 outcomes above, we can see that two of them are female (XX) and two of them are male (XY); there is a 50% chance of the offspring being female and a 50% chance of the offspring being male.



- ▶ **Fur colour** in **mice** is controlled by a **single gene**. The allele that codes for **white fur** is **recessive** and represented by a lowercase b in this example. The allele that codes for **grey fur** is **dominant** and is represented by an uppercase B in this example.

1. When a **grey homozygous dominant** mouse (**BB**) breeds with a **white homozygous recessive** mouse (**bb**), all of their offspring will be **grey** and will be **heterozygous (Bb)**.

All offspring in the **punnet square** are **grey** so the probability of an offspring being grey is 100% and probability of it being white is 0%. And they are all normal.

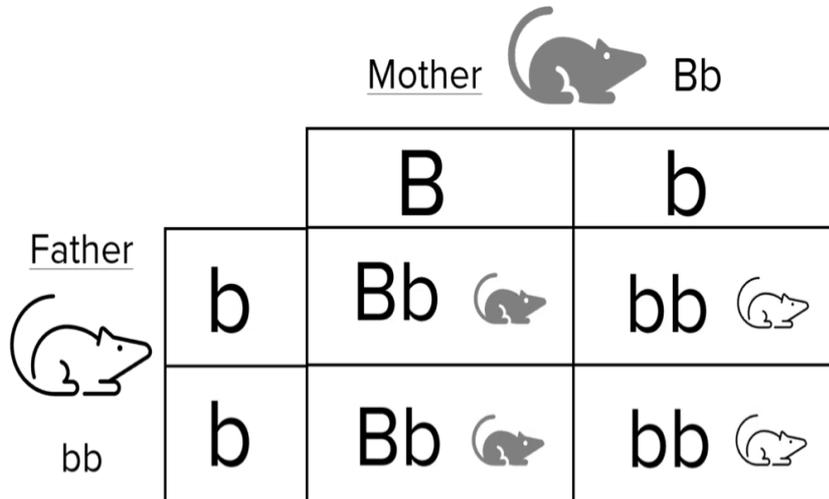
		Mother  BB	
		B	B
Father  bb	b	Bb 	Bb 
	b	Bb 	Bb 

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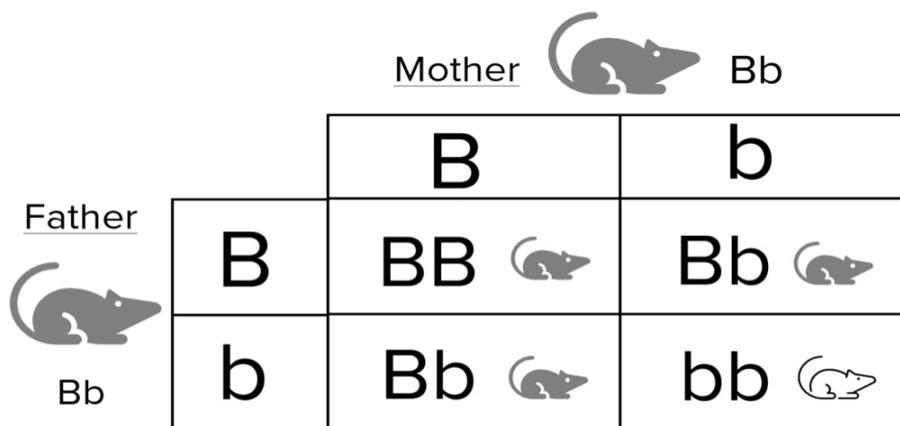
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2. When a **grey heterozygous (Bb)** mouse breeds with a **white homozygous recessive (bb)** mouse, there is an equal chance that the offspring will be **grey heterozygous (Bb)** and being **white homozygous (bb)**.



22 out of 44 boxes have white offspring, 22 out of 44 boxes have grey offspring. There is a 50% chance of the offspring being **grey** and a 50% chance of the offspring being **white**. The ratio of white to grey mice is 1:1.

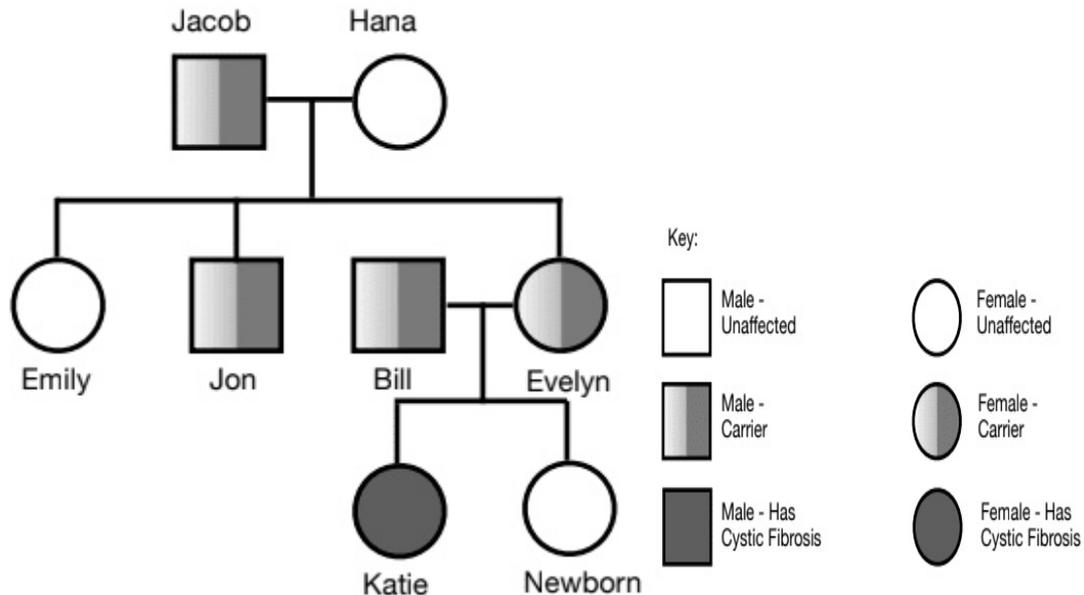
3. When a **grey heterozygous (Bb)** mouse breeds with another **grey heterozygous (Bb)** mouse, there is 25% chance of the offspring being **homozygous grey (BB)**, a 25% chance of the offspring being **homozygous white (bb)** and a **50%** of the offspring being **grey and heterozygous (Bb)**.



► **33 out of the 44 boxes** show grey offspring and 11 out of the 44 boxes show white offspring. Overall, the chance having of **grey** offspring is 75% and the chance of having **white** offspring is 25%. The ratio of white to grey mice is 1:3.

Family Pedigrees

- ▶ A **pedigree** is a family tree that shows how genetic disorders are passed down the family.
- ▶ We can understand pedigrees by looking at cystic fibrosis in a family as an example of cystic Fibrosis



1. **Cystic fibrosis is a recessive disease.** So if a person is homozygous recessive, they will have the disease.
2. **If they are heterozygous,** they will be carriers of the disease. This means that they don't have the disease but it can still be passed down the family like Ff gene.
3. **If they are homozygous dominant,** they won't have the disease or be carriers. Like FF.

Punnet square or genetic diagram of Cystic fibrosis

- ▶ To find what is the probability of new born we draw a punnet square of this. This genetic diagram below shows the possible outcomes when both parents are heterozygous for the faulty allele. (Mother and father both have one F and one f allele)
- ▶ There is a one in four chance of the offspring being homozygous for the faulty allele, and so having cystic fibrosis.

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f is the cystic
fibrosis allele

		mother	
		F	f
father	F	FF	Ff
	f	Ff	ff