



Chromosome deletions and microdeletions

This communication aid has been produced for clinicians to help support and guide conversations about chromosome deletions and microdeletions with their patients.

Chromosomes explained

We all have over 20,000 genes, which provide instructions for how our body works. Our genes are packaged into structures called **chromosomes**.

People usually have 23 pairs of chromosomes: 22 pairs of **autosomes** and one pair of **sex chromosomes** (XX or XY). One chromosome from each pair is inherited from each parent. Our sex chromosomes are called X or Y, and usually determine sex assigned at birth.

Chromosomes have a short arm, known as the p arm, and a long arm, known as the q arm. The p arm and q arm are joined together by a centromere.

You can learn more about the basics of our genome in the VCA: Genes, chromosomes and DNA (available via the QR code, below).

Key terms

Genome: The complete set of genetic information found inside a cell.

Chromosome: Packages of DNA that are found in our cells.

Autosome: A chromosome that is not a sex chromosome (named 1–22).

Sex chromosome: A chromosome containing genes that determine the sex a person is born as (named X or Y).

What are chromosome deletions and microdeletions?

A deletion is a loss of DNA from a chromosome. Deletions can be large or small. Large deletions can be seen under a microscope.

Deletions that are too small to be seen under a microscope are known as microdeletions.

A deletion may include a part of a gene, a whole gene or multiple genes. Deletions are usually described based on the chromosome affected and the location of the deletion on the chromosome.

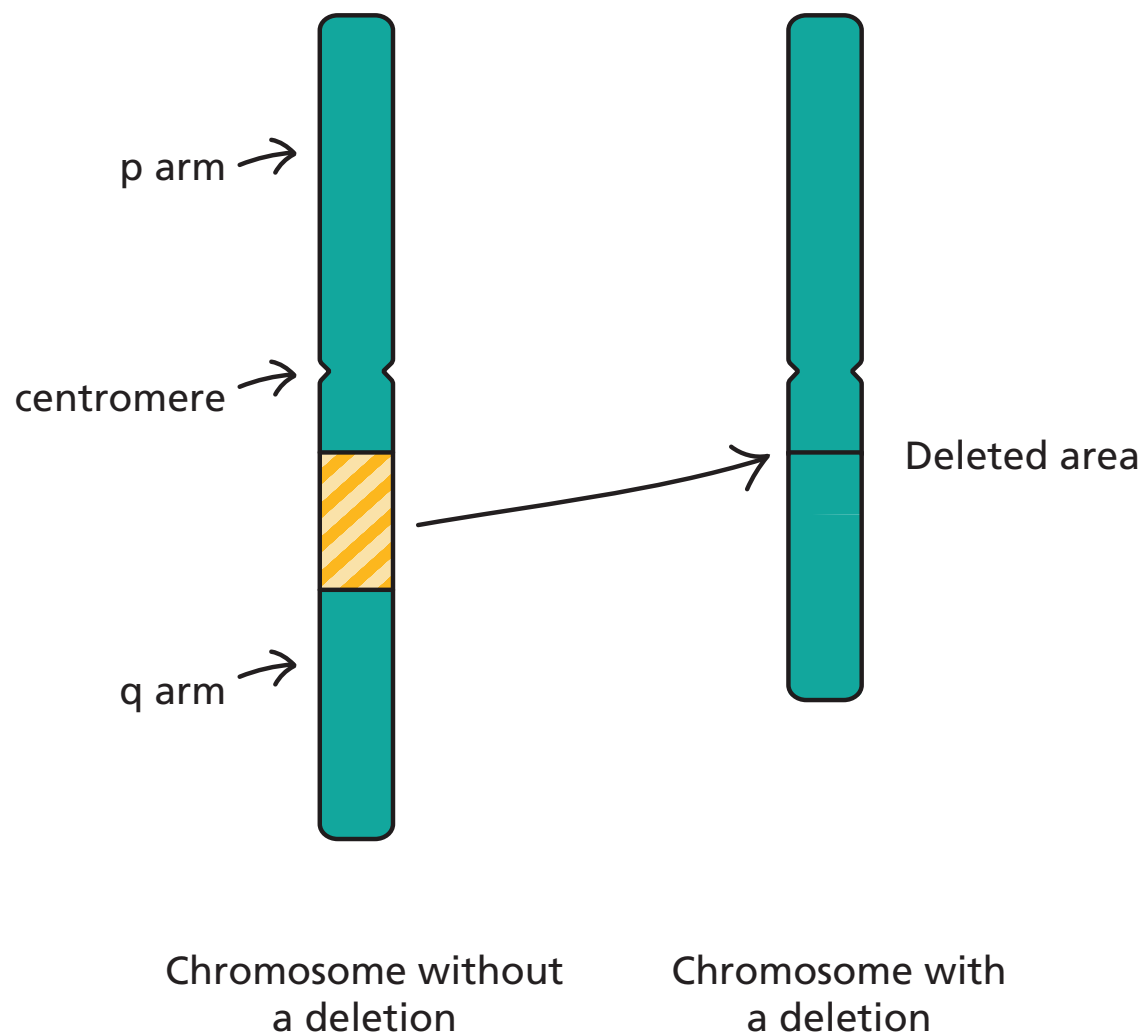
Some deletions may not have any effect on a person's health. Some deletions may cause a genetic condition.





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