



# Chromosome duplications and microduplications

This communication aid has been produced for clinicians to help support and guide conversations about chromosome duplications and microduplications 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 duplications and microduplications?

A duplication is a gain of DNA, where a section of chromosome has an extra copy. Duplications can be large or small. Large duplications can be seen under a microscope. Duplications that are too small to be seen under a microscope are known as microduplications.

A duplication may include a part of a gene, a whole gene or multiple genes.

Duplications are usually described based on the chromosome affected and the location of the duplication on the chromosome.

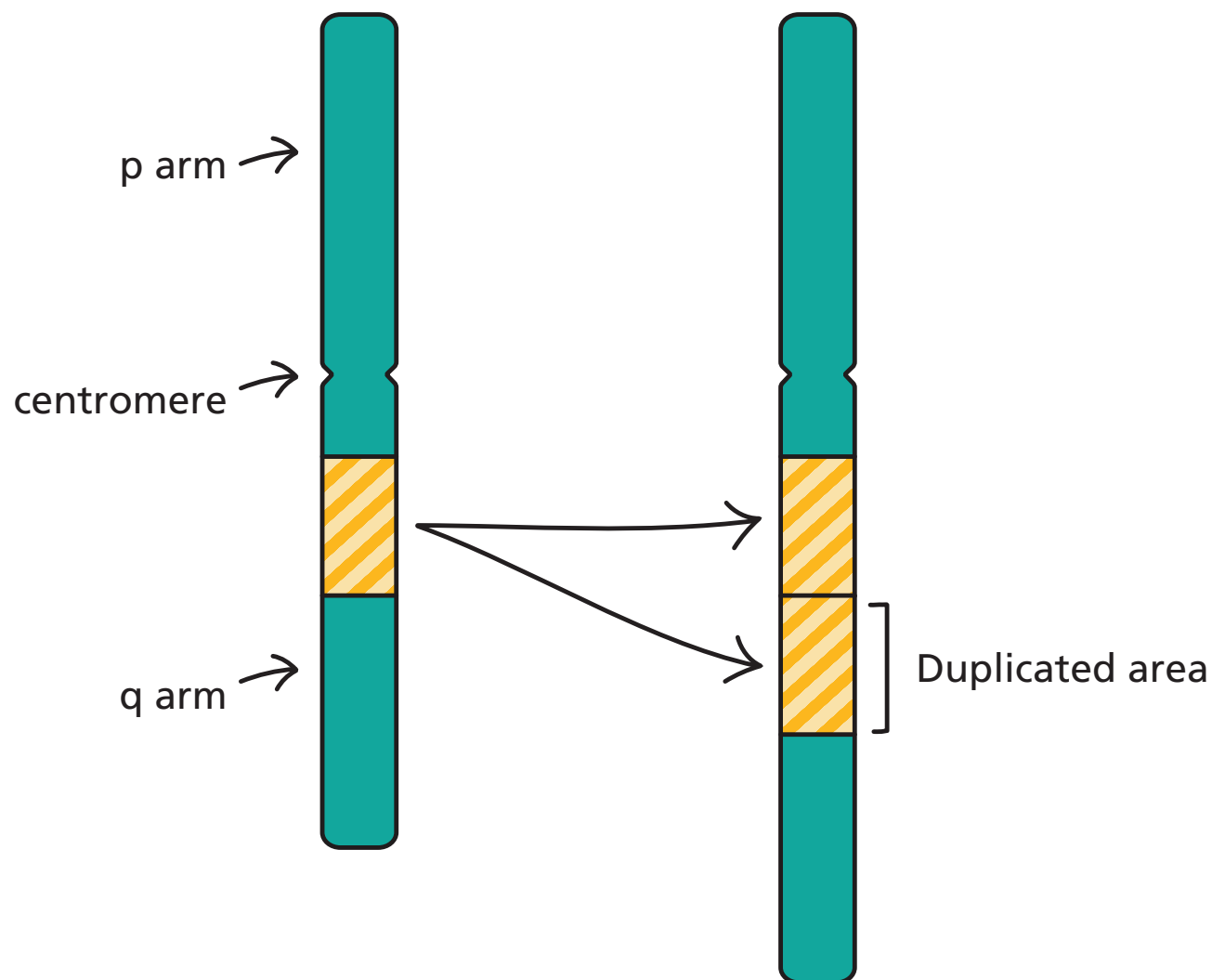
Some duplications may not have any effect on a person's health. Some duplications may cause a genetic condition.





# Chromosome duplications and microduplications

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



Please scan to leave feedback  
about this resource



Last reviewed: 4 November 2025