

Mosaicism is a term that describes when two different types of cells can be found in one person. We know from Genetics 101 that each cell of our body contains chromosomes that act as an instruction manual telling our body how to grow, develop, and function. We usually have 23 pairs of chromosomes, one set coming from mom and the other set from dad, which gives us a total of 46 chromosomes. The body is very particular in the amount of this information that we have, and too many or too few of these instructions can change how things work in the body.

In the simplest form of mosaicism, an individual may have 46 chromosomes in some cells of their body and 45 chromosomes (missing a chromosome) in some cells of their body. For example, most females have two X chromosomes, while most males have one X and one Y chromosome. However, there is a genetic condition called **Turner syndrome** in which females only have one X chromosome (they do not have a second X chromosome). Some females who have <u>Turner syndrome</u> actually have mosaic <u>Turner syndrome</u>, meaning that some cells in their body have two X chromosomes (not <u>Turner syndrome</u>), while other cells in their body only have one X chromosome (<u>Turner syndrome</u>). Mosaicism can happen in the case of a whole chromosome as noted above or can happen for a single gene, or single instruction.

People who have mosaic genetic conditions, such as <u>Turner syndrome</u>, do not generally have as many of the health concerns as someone who has the genetic condition in every single cell of their body. It can also be difficult to determine if someone has mosaicism, and if they do, which cells in the body may be affected. Even if it is determined that someone is mosaic, there is not an easy way to determine what percentage of their cells are 'normal' and what percentage of their cells are 'abnormal', and where in the body each type of cell is.