

Mosaicism

Genetic Basis of Mosaicism

The term “mosaicism” describes a situation in which different cells in the same individual have different numbers or arrangements of chromosomes. It is called “mosaicism” because, in a way, the cells of the body are similar to the tiles of a mosaic. In a mosaic piece of art, each tile is different. They have different shapes and colors. The tiles are fitted together to make a whole picture.



If a person has mosaicism, their cells are like the tiles of a mosaic. Taken together, the different tiles of the mosaic form the whole picture, similar to the way the cells form the whole body. Just as the tiles of a mosaic have different shapes and colors, the cells of the body have different numbers or arrangements of chromosomes. For example, if an individual has mosaic trisomy 18, this means that some of the cells have three copies of chromosome 18 while other cells have two copies of chromosome 18.

Diagnosis of Mosaicism

Mosaicism can be diagnosed in different ways. Sometimes, different cells in the blood have different chromosome make-ups. For these individuals, a blood test may be able to detect mosaicism. In other individuals, it is more difficult to diagnose mosaicism. For example, a blood test would not be able to diagnose mosaicism if the chromosome change is only located in the skin cells. In cases like this, additional testing may be needed to detect mosaicism. Usually, this would involve examination of other tissues of the body, such as the skin. A geneticist can help determine whether this type of additional testing is appropriate.

Characteristics of Mosaicism

In general, individuals with mosaicism have similar medical and developmental concerns to those with a non-mosaic form of the same chromosome condition. For example, an individual with mosaic tetrasomy 18p may have many of the same features as someone with non-mosaic tetrasomy 18p. However, because individuals with mosaicism have fewer cells carrying the chromosome change, they tend to be less severely affected. It is very important to understand that this is not always the case.

Unfortunately, it is impossible to predict exactly how mosaicism will affect an individual at this point in time. This is because we cannot know precisely which cells carry the chromosome change and which cells do not.

Germline Mosaicism

The term “germline mosaicism” refers to a particular type of mosaicism that only affects one group of cells. The germline cells include eggs and sperm. In some individuals that have had a child with a chromosome change, there is mosaicism in these cells. This means that some eggs or sperm have a normal chromosomal make-up while others carry a chromosome change. If one of the eggs or sperm with a chromosome change is used in fertilization, the child may have a chromosome abnormality.

Unfortunately, it is impossible to determine whether an individual has germline mosaicism. To do so would mean that every egg or sperm would have to be tested. However, it is important to realize that germline mosaicism is a very rare occurrence in a parent who has had a child with a chromosome change.

Family Planning and Genetic Counseling

If a person has mosaicism (including germline mosaicism), it is impossible to estimate the precise likelihood that he or she would have a child with a chromosome change. However, there are multiple options for preimplantation and prenatal diagnosis, if a family is interested in exploring them. Preimplantation genetic diagnosis involves screening embryos for genetic conditions before implanting the embryos in the mother’s uterus. Prenatal diagnosis involves testing the fetus through one of several different techniques, such as chorionic villus sampling or amniocentesis. To learn more about these options, we recommend contacting a genetics provider.

For Additional Information

The information provided here is general information based on the literature as well as the experiences in the Chromosome 18 Clinical Research Center. If you have questions or concerns, you may find it helpful to talk with a clinical geneticist or genetic counselor. You can locate a genetics provider at one of these sites:

- [GeneTests Clinic Directory](#)
- [National Society of Genetic Counselors](#)