Mosaic Down Syndrome

WHAT IS MOSAIC DOWN SYNDROME?
Mosaic Down syndrome happens when an extra copy of chromosome 21 is present in some, but not all, of the body’s cells.

- Chromosomes contain all of the genetic information that tells our body how to grow and function.
- Most people have 46 total chromosomes (23 pairs) in every cell in their body. One chromosome of each pair comes from the father, and one chromosome of each pair comes from the mother.
- An extra copy of chromosome 21 causes the differences we see in people with Down syndrome.
- About 1 to 2% of people with Down syndrome have mosaic Down syndrome.

WHAT CAUSES MOSAIC DOWN SYNDROME?
Mosaic Down syndrome is caused by a random event shortly after the egg and sperm join together.

- When cells are dividing, some cells receive an extra copy of chromosome 21. Others do not.
- People with mosaic Down syndrome have some cells with two and some cells with three copies of chromosome 21. There is no way of knowing which, or how many, cells have two or three copies of chromosome 21.

WHAT DOES MOSAICISM MEAN FOR MY CHILD?
- People with mosaic Down syndrome share many of the same health issues and learning problems with people who have other types of Down syndrome.
- The specific amount of extra genes generally does not help us predict how a child will develop.

WHAT DO MOSAIC CHROMOSOMES LOOK LIKE?
Some cells have two copies of chromosome 21, while other cells have three copies of chromosome 21. The diagrams below show what two different kinds of cells look like in the same person.

COULD I HAVE ANOTHER BABY WITH DOWN SYNDROME?
Mosaic Down syndrome happens by chance. It is not passed down by parents.

- Anyone of any age can have a baby with Down syndrome.
- The chance of having a second baby with Down syndrome is approximately 1%. The chance may be slightly higher depending on the age of the mother.
- A genetic counselor or doctor who is a Down syndrome expert would be happy to talk about this with you before any future pregnancies.