Translocation Down Syndrome

**WHAT IS TRANSLOCATION DOWN SYNDROME?**
Translocation Down syndrome happens when an extra copy of chromosome 21 is attached to another chromosome. This is then present in all cells of the body.

- 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 3 – 4% of people with Down syndrome have translocation Down syndrome.

**WHAT DOES TRANSLOCATION DOWN SYNDROME LOOK LIKE?**
In Translocation Down syndrome, an extra copy of chromosome 21 is attached to a different chromosome. For example, when an extra chromosome 21 is attached to chromosome 14, it looks like the picture below.

**WHAT CAUSES TRANSLOCATION DOWN SYNDROME?**
Translocation Down syndrome can happen one of two ways:

- It can be caused by extra genes in the egg or sperm of one of the parents.
- It can happen by chance.

**WHAT IS BALANCED TRANSLOCATION?**
If the extra copy of chromosome 21 is inherited from a parent in the egg or sperm, it means that the parents “carry” this type of Down syndrome. These parents are said to have a “balanced translocation.” Balanced translocation happens when the correct amount of genetic material is present in the wrong location.

Balanced Translocation
DO PEOPLE WITH BALANCED TRANSLOCATION HAVE SIGNS OF DOWN SYNDROME?
People with balanced translocations do not have any features of Down syndrome. However, it is possible that they may have trouble getting pregnant.

- Even though carriers have the right amount of genes, the egg or sperm may have either too much or too few genes. This may cause an unexpected miscarriage.

WHAT DOES TRANSLOCATION DOWN SYNDROME MEAN FOR MY CHILD?
- People with translocation Down syndrome still share the same health issues and learning problems with people with other types of Down syndrome.
- Without doing a blood test, it is not possible to tell the difference between people with translocation Down syndrome and people with other types of Down syndrome.
- All people with Down syndrome have some learning problems. The specific location of extra genes generally does not help us predict how a child will develop.

COULD I HAVE ANOTHER BABY WITH DOWN SYNDROME?
Yes. Translocation Down syndrome is the only type of Down syndrome that can be passed down from a parent who does not have features of Down syndrome.

- If a parent has balanced translocation, there is an up to 15% chance of having another child with Down syndrome.
- A genetic counselor or doctor who is a Down syndrome expert would be happy to discuss this with you before any future pregnancies.
- These counselors or doctors can also discuss which other family members have a chance of carrying the balanced translocation, such as brothers or sisters.