

Trisomy 21 Down Syndrome

WHAT IS TRISOMY 21?

Trisomy 21 Down syndrome happens when an extra copy of chromosome 21 is present in all cells of the body.

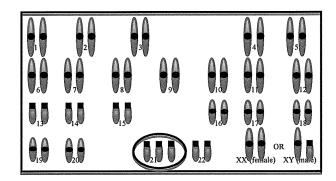
- Chromosomes contain all of the genetic information that tell our body how to grow
- Most people have 46 total chromosomes (23 pairs) in every cell in their body. One chromosome of each pair comes from the father, and one chromosome of each pair comes from the mother.
- An extra copy of chromosome 21 causes the differences we see in people with Down syndrome.
- About 95% of people with Down syndrome have trisomy 21 Down syndrome.

WHAT CAUSES TRISOMY 21?

- The egg and sperm typically contain one copy of every chromosome. When they combine, they produce a full set of chromosomal material.
- Sometimes, chromosomes do not divide properly. For example, two copies of chromosome 21 may "stick" together in an egg. When that egg combines with the sperm's one copy of chromosome 21, there will then be three copies of chromosome 21.
- In the majority of cases, the egg passes on the extra chromosome. But there are some cases where the sperm passes on the extra chromosome.
- It is important to know that nothing a mother or father ever did would cause Down syndrome. The extra chromosome is passed on by chance.
- An extra chromosome 21 is present in every cell.

WHAT DOES TRISOMY 21 LOOK LIKE?

A person with trisomy 21 will have three copies of chromosome 21 in every cell of the body. A typical person has just two copies of chromosome 21. The picture below shows what the chromosomes look like in one cell of a person with trisomy 21.



COULD I HAVE ANOTHER BABY WITH DOWN SYNDROME?

Trisomy 21 Down syndrome happens by chance. It 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.

If you have any feedback on this document, please contact mghfcpatienteducation@partners.org.

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