

# Down Syndrome: The Role of Genetics and Chromosomal Abnormalities

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## DESCRIPTION

Down syndrome or trisomy 21, is a genetic condition that leads to intellectual disabilities and a range of physical health issues. It occurs when a person inherits an extra copy of chromosome 21, resulting in three chromosomes instead of the usual two. This additional chromosome interferes with normal development, giving rise to the distinctive traits and challenges associated with the condition. The causes of down syndrome are deeply rooted in genetics, specifically in chromosomal abnormalities that affect cell division during reproduction. These causes important for providing better care, early intervention and support to those with the condition. This article examines the role of genetics and chromosomal abnormalities in the development of down syndrome.

## Genetics and chromosomal abnormalities in down syndrome

These chromosomes carry the genetic information that determines an individual's traits, from physical characteristics to how the body functions. However, in individuals with down syndrome, the presence of an extra chromosome 21 causes a series of genetic changes that lead to the condition's physical and developmental traits

**Trisomy 21 (Nondisjunction):** This occurs due to a cell division error known as nondisjunction, which happens during the formation of egg or sperm cells. As a result, instead of receiving one copy of chromosome 21 from each parent, the child ends up with two copies of chromosome 21 from one parent and only one from the other. This leads to a total of three chromosome 21s and the additional genetic material interferes with normal development, causing the characteristic features of down syndrome. The error in cell division can occur in either the egg or sperm, but maternal age is known to increase the risk of nondisjunction, with older mothers being more likely to have children with down syndrome.

**Translocation down syndrome:** Translocation down syndrome accounts for around 3% of cases and involves a rearrangement of chromosome 21. This means the individual still has two copies of chromosome 21, but one is attached to another chromosome,

causing the same genetic effects as trisomy 21. Unlike nondisjunction, translocation can be inherited, meaning that one of the parents might carry a balanced translocation, which does not cause symptoms in them but can lead to down syndrome in their children. Genetic testing can identify this type of down syndrome and determine whether a parent is a carrier.

**Mosaic down syndrome:** Mosaic down syndrome is a rarer form, occurring in about 2% of cases. This condition is characterized by a mix of cells, some having the typical two copies of chromosome 21, while others have three copies. This mosaic pattern results from an error in cell division after fertilization, meaning that only some of the individual's cells have the extra chromosome. The severity of symptoms in mosaic down syndrome can vary, as those with more normal cells may experience fewer physical and developmental challenges compared to those with more cells affected by the chromosomal abnormality.

## Genetic factors in the development of down syndrome

The main cause of down syndrome is the presence of an additional chromosome 21, which disrupts the normal balance of gene activity. The additional genetic material from the third chromosome 21 affects the development of the brain and other organs, leading to intellectual disabilities and the physical features commonly seen in individuals with down syndrome, such as a flat facial profile, almond-shaped eyes and a short stature.

The extra genetic material causes overexpression of certain genes, which can disrupt normal cellular processes. The overexpression of genes on chromosome 21 affects various pathways in the body, such as those responsible for growth, brain development and cell division. This overexpression contributes to developmental delays and intellectual disabilities, as well as other medical conditions, including heart defects, gastrointestinal problems and hearing or vision impairment, which are more common in individuals with down syndrome.

Additionally, the role of genetics in down syndrome extends beyond the chromosomal abnormalities themselves. Genetic

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factors, including family history and the mother's age, can affect the chances of the condition occurring. For instance, older mothers, especially those over the age of 35, have a higher chance of having a child with down syndrome due to the increased likelihood of errors in cell division during egg formation.

## CONCLUSION

The three main types of down syndrome trisomy 21, translocation and mosaicism each have different genetic

mechanisms but all result in similar outcomes in terms of physical and cognitive development. Insight of genetic basis of down syndrome is essential for both diagnosis and treatment, as it provides insight into the underlying causes of the condition and informs early intervention strategies. While there is no cure for down syndrome, analyzing genetic therapies and early intervention programs continues to improve the quality of life for those affected. By gaining a deeper importance of the role genetics plays in down syndrome, we can better support individuals with the condition, help their families and promote inclusion in society.