What Causes Down Syndrome?

The set of birth characteristics known as Down syndrome is usually caused by an error in cell division, typically called nondisjunction.

However, two other types of chromosomal abnormalities, mosacism and translocation also occur, but to a much lesser extent. Regardless of the type of Down syndrome which a person may have, all people with Down syndrome have an extra, critical portion of the number 21 chromosome present in all, or some of their cells.

Nondisjunction:

Nondisjunction is a faulty cell division which results in the development of an embryo with three number 21 chromosomes instead of two. Prior to, or, at conception, a pair of number 21 chromosomes, in either the sperm or the egg, fail to separate. As the embryo develops, the extra chromosome is replicated in every cell of the body. This faulty cell division is responsible for 95 percent of all cases of Down syndrome.

This additional genetic material alters the course of development and causes the characteristics associated with the syndrome.

Why this error in cell division occurs is currently unknown, although it does seem to be related to advancing maternal age. Many people are surprised to find that 80 % of children with Down syndrome are born to women under 35 years of age. This occurs because younger women have higher fertility rates. However this does not contradict the fact that the incidence of births of children with Down syndrome increases with maternal age.

Despite years of research the cause/s of nondisjunction is still unknown. There seems to be no connection between any type of Down syndrome and parents' activities before or during pregnancy.

Mosacism

Mosaicism occurs when there is a mixture of two types of cells, some containing 46 chromosomes and some containing 47.

Those cells with 47 chromosomes contain an extra 21st chromosome. Because of the random pattern of the cells the term mosacism is used. This particular form of Down syndrome is rare, making up only 2% of all cases.

Some research has indicated that individuals with mosaic Down syndrome are less affected by the disability than those with typical trisomy 21.

Translocation

Translocation is another chromosomal problem which occurs in three to four percent of people with Down syndrome. Part of the number 21 chromosome breaks off during cell division and attaches to another chromosome. While the total number of chromosomes in the cell remains 46, the presence of an extra part of the number 21 chromosome causes the features of Down syndrome. As with common trisomy 21, translocation occurs either prior to or at conception.

Unlike nondisjunction, maternal age is not linked to the risk of translocation. Most cases are random, chance events. However, in about one-third of cases, one parent is a carrier of a translocated chromosome. For this reason, the risk of recurrence in subsequent births is higher than that of nondisjunction.

For more information contact:

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