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Commentary

Genetic Challenges of Down Syndrome

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DESCRIPTION

Down syndrome also known as trisomy 21, is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21. It is usually associated with physical growth delays, mild to moderate intellectual disability, and characteristic facial features. The average IQ of a young adult with Down syndrome is 50, equivalent to the mental ability of an eight- or nine-year-old child, but this can vary widely. They are also more likely to have gastric reflux illness, celiac disease, hypothyroidism, hearing and vision issues, leukemia, and Alzheimer's disease. Down syndrome is caused by having three copies of chromosome 21 (also known as trisomy 21) instead of the usual two copies, and it is not usually inherited. The treatment focuses on the individual's specific symptoms. The particular genes that cause the condition are still being researched in order to identify more effective treatments. Medical issues that may arise in people with Down syndrome include:

Congenital hypothyroidism

Reduced thyroid hormone production in a newborn is known as neonatal hypothyroidism. Thyroid hormone is only produced in a small percentage of cases. Congenital hypothyroidism is another name for the disorder. A condition that is present from birth is referred to as congenital.

Hearing loss

It is defined as the lack or decreased ability to hear sound. Hearing loss can damage one or both ears. A mechanical problem, such as damage to the tiny bones in the ear, or a problem sending a signal to the brain indicating that a sound has been heard, can cause hearing loss. Hearing loss can be present at birth or develop over time, and it can be caused by a variety of reasons including genetics, infectious infections, trauma, certain drugs, long-term loud noise exposure, and ageing.

Congenital heart defect

It is an abnormality in the structure or function of the heart that is present at birth. The chambers of the heart, the valves linking the chambers, or the blood flow in the heart can all be affected by this abnormality. To address the defect, a cardiologist may recommend close monitoring, medicines, or surgical procedures.

Seizures

A seizure is a type of aberrant electrical activity in the brain that causes uncontrollable body movements, behavioral changes, and occasionally even loss of consciousness. Some seizures have milder signs, and they may go unnoticed. Injuries, illnesses, high fevers, or an underlying ailment can all trigger seizures. Epilepsy is a condition in which an individual suffers from epileptic seizures.

Decreased muscle tone (hypotonia)

Muscle tone that is reduced or absent. This can make the person appear "floppy", and they may be unable to move as well as they should. Hypotonia may have a hereditary basis in some circumstances; Down syndrome can be caused by one of three genetic factors.

Trisomy 21 is a genetic disorder

The most common cause of Down syndrome is an extra chromosome 21 in all of the affected person's cells. The chromosome 21 pair fails to separate during the development of an egg (or sperm) in certain circumstances, which is known as non-disjunction. When a normal sperm with one copy of chromosome 21 combines with an egg with two copies of chromosome 21, the resulting embryo has three copies of chromosome 21 instead of the typical two. The extra chromosome is then replicated in every cell of the baby's body, resulting in Down syndrome's symptoms. The origin of nondisjunction is unknown, although studies show that it occurs more frequently as women get older. Nothing in the environment or what parents do (or don't do) before or during pregnancy is known to induce nondisjunction.

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Mosaic trisomy 21 is a type of mosaic trisomy

Only some of a person's cells have an additional chromosome 21 in roughly 1-2 percent of cases; this is known as "mosaic trisomy 21". In this case, the fertilized egg may have the correct number of chromosomes, but certain cells "acquire" an additional chromosome 21 due to a cell division error early in the embryo's development. Mosaic trisomy 21 is characterized by 46 chromosomes in some cells and 47 chromosomes (including the additional chromosome 21) in others. Mosaic trisomy 21 can have a wide range of symptoms and severity.

Trisomy 21 (translocation)

Cells in people with Down syndrome contain 46 chromosomes, however there is additional chromosome 21 material connected (translocate) to another chromosome in about 3-4 percent of cases. There may be an increased risk of Down syndrome in future pregnancies for parents of a child with Down syndrome due to a translocation. This is due to the possibility that one of the two parents is a balanced translocation carrier. However, not all people with translocation trisomy 21 have a translocation in their parents. All people with Down syndrome have an additional, important piece of chromosome 21 present in all or some of their cells, regardless of the form of Down syndrome they have. The additional genetic material affects the usual path of development, resulting in the Down syndrome symptoms.

Diagnosis

Diagnostic tests can detect whether a foetus has Down syndrome with certainty during pregnancy, but they are intrusive and involve a risk of miscarriage. Chorionic villus sampling in the first trimester and amniocentesis in the second trimester are two examples of diagnostic tests. A small sample of genetic material is taken from the amniotic fluid or placenta during these procedures, and the foetus chromosomes are subsequently examined in a lab. Non-Invasive Prenatal Testing (NIPT) has become available to women who are at a higher risk of having a baby with Down syndrome in recent years. The NIPT test looks for DNA from the foetus in the mother's bloodstream. Women with a positive NIPT result, on the other hand, should undertake invasive diagnostic testing to confirm the finding.