DOWN'S

SYNDROME

Complied by

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IMPORTANT

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Introduction

Down syndrome, also known as trisomy 21, is a chromosomal disorder caused by an error in cell division that results in an extra 21st chromosome. It is named after John Langdon Down, the British physician who described the syndrome in 1866. The condition was clinically described earlier in the 19th century by Jean Etienne Dominique Esquirol in 1838 and Edouard Seguin in 1844. Down syndrome was identified as a chromosome 21 trisomy by Dr. Jérôme Lejeune in 1959.

The incidence of Down syndrome is estimated at 4.6 per 10,000 births. Down syndrome occurs in all human populations, and analogous conditions have been found in other species such as chimpanzees.

The condition leads to impairments in both cognitive ability and physical growth that range from mild to moderate developmental disabilities. Through a series of screenings and tests, Down syndrome can be detected before and after a baby is born.

The only factor known to affect the probability of having a baby with Down syndrome is maternal age. That is, less than one in 1,000 pregnancies for mothers less than 30 years of age results in a baby with Down syndrome. For mothers who are 44 years of age, about 1 in 35 pregnancies results in a baby with Down syndrome. Because younger women generally have more children, about 75 - 80% of children with Down syndrome are born to younger women.

Within every cell in the human body there is a nucleus, where genetic material is stored in genes. Genes carry the codes responsible for all of our inherited traits and are grouped along rod-like structures called chromosomes. Normally, the nucleus of each cell contains 23 pairs of chromosomes, half of which are inherited from each parent.

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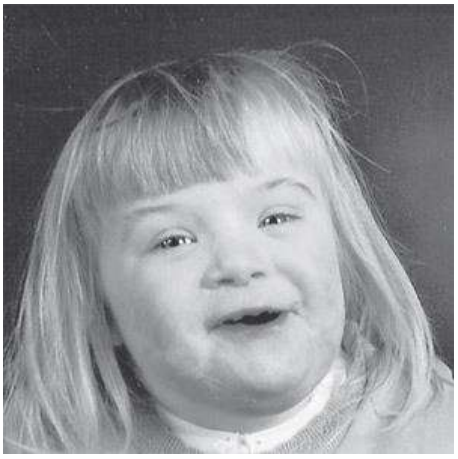
Cell Division Error

Down syndrome is usually caused by an error in cell division called "nondisjunction." Nondisjunction results in an embryo with three copies of chromosome 21 instead of the usual two. Prior to or at conception, a pair of 21st chromosomes in either the sperm or the egg fails to separate. As the embryo develops, the extra chromosome is replicated in every cell of the body. This type of Down syndrome, which accounts for 95% of cases, is called Trisomy 21.



The two other types of Down syndrome are called mosaicism and translocation. Mosaicism occurs when nondisjunction of chromosome 21 takes place in one-but not all-of the initial cell divisions after fertilization. When this occurs, there is a mixture of two types of cells, some containing the usual 46 chromosomes and others containing 47. Those cells with 47 chromosomes contain an extra chromosome 21. Mosaicism accounts for about 1% of all cases of Down syndrome. Research has indicated that individuals with mosaic Down syndrome may have fewer characteristics of Down syndrome than those with other types of Down syndrome. However, broad generalizations are not possible due to the wide range of abilities people with Down syndrome possess.

Translocation accounts for about 4% of all cases of Down syndrome. In translocation, part of chromosome 21 breaks off during cell division and attaches to another chromosome, typically chromosome 14. While the total number of chromosomes in the cells remain 46, the presence of an extra part of chromosome 21 causes the characteristics of Down syndrome.

Regardless of the type of Down syndrome a person may have, all people with Down syndrome have an extra, critical portion of chromosome 21 present in all or some of their cells. This additional genetic material alters the course of development and causes the characteristics associated with Down syndrome.

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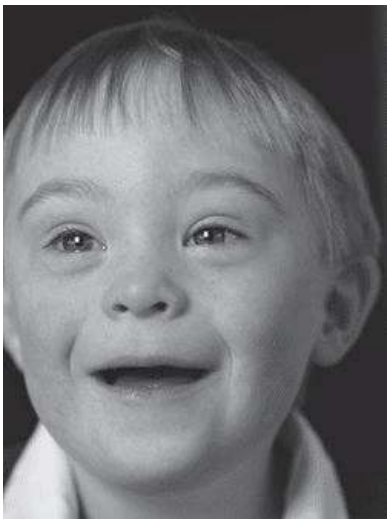
Cause

The cause of nondisjunction is currently unknown, but research has shown that it increases in frequency as a woman ages. However, due to higher birth rates in younger women, 80% of children with Down syndrome are born to women under 35 years of age. There is no definitive scientific research that indicates that Down syndrome is caused by environmental factors or the parents' activities before or during pregnancy.

(Above - Characteristics of Down's Syndrome)

Once a woman has given birth to a baby with Trisomy 21, it is estimated that her chances of having another baby with Trisomy 21 is 1% greater than her chances by age alone.



The age of the mother does not seem to be linked to the risk of translocation. Most cases are sporadic-that is, chance events. However, in about one third of cases, one parent is a carrier of a translocated chromosome. The risk of recurrence of translocation is about 3% if the father is the carrier and 10-15% if the mother is the carrier. Genetic counseling can determine the origin of translocation.

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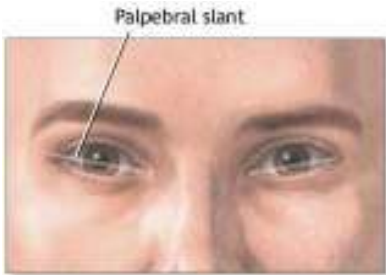
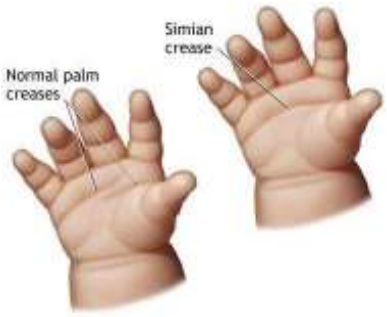
Characteristics

Down syndrome often has distinct physical characteristics, unique health issues, and variability in cognitive development.

Physical characteristics include:



- Eyes - White spots on the iris (Brushfield Spots - Picture Right)
- Eyes - Oblique fissures, epicanthic skin folds on the inner corner
- Eyes - Strabismus (Strabismus allows bringing the gaze of each eye to a different point in space)
- Eyes- Upward slant (Palebral Slant - Picture Right)
- Low muscle tone
- Congenital heart disease
- Small stature and short neck
- Shortened hands
- Flat nasal bridge
- Protruding possible large tongue
- Epicanthal fold (Mongoloid eye)
- Single, deep creases across the centre of the palm
- Simian (single palmer) crease (Picture Right)
- A single flexion furrow of the fifth finger
- Eyes with upward (palpebral) slant
- Small Teeth
- Large space between large and second toe (Picture Below Right)



Additionally, individuals with Down syndrome usually have cognitive development profiles indicative of mild to moderate mental retardation. However, cognitive development in children with Down syndrome is quite variable.

Children with Down syndrome often have a speech delay and require speech therapy to assist with expressive language. In addition, fine motor skills are delayed and tend to lag behind gross motor skills. Children with Down syndrome may not walk until age 4, but some will walk at age 2.

Although many with the condition experience developmental delays, it is not uncommon for those with Down syndrome to attend school and to become active, productive members in the community.

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Screening and Diagnosis

Screening tests include:

- Nuchal translucency testing (at 11 to 14 weeks) - an ultrasound that measures clear space in folds of tissue behind the neck of a developing baby.

- Triple screen or quadruple screen (at 15 to 18 weeks) - measures the quantities of normal substances in the mother's blood.
- Integrated screen - combines first trimester screening tests (with or without nuchal translucency) and blood tests with second trimester quadruple screen.
- Genetic ultrasound (at 18 to 20 weeks) - Detailed ultrasound combined with blood test results.

Diagnostic tests include:

- Chorionic villus sampling (at 8 to 12 weeks) - analysis of a tiny sample of placenta obtained from a needle inserted into the cervix or the abdomen.
- Amniocentesis (at 15 to 20 weeks) - analysis of a small amount of amniotic fluid obtained from a needle inserted into the abdomen.
- Percutaneous umbilical blood sampling (after 20 weeks) - analysis of a small sample of blood from the umbilical cord obtained from a needle inserted into the abdomen.

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Inspection

The condition can also be diagnosed after a baby is born by inspecting the infant's physical characteristics as well as blood and tissue samples that are stained to show chromosomes grouped by size, number, and shape.

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