DOWN SYNDROME FACTS

NADS

National Association for Down Syndrome

The mission of NADS is to ensure that all persons with Down syndrome have the opportunity to achieve their potential in all aspects of community life.

DOWN SYNDROME

Down syndrome is a genetic condition that causes delays in physical and intellectual development. It occurs in approximately one in every 800 live births. Individuals with Down syndrome have 47 chromosomes instead of the usual 46. It is the most frequently occurring chromosomal disorder. Down syndrome is not related to race, nationality, religion or socioeconomic status. The most important fact to know about individuals with Down syndrome is that they are more like others than they are different.

DIAGNOSIS

Down syndrome is usually identified at birth or shortly thereafter. Initially the diagnosis is based on physical characteristics that are commonly seen in babies with Down syndrome. These include low muscle tone, a single crease across the palm of the hand, a slightly flattened facial profile and an upward slant to the eyes. The diagnosis must be confirmed by a chromosome study (karyotype). A karyotype provides a visual display of the chromosomes grouped by their size, number and shape. Chromosomes may be studied by examining blood or tissue cells.

CAUSE

Down syndrome is usually caused by an error in cell division called nondisjunction. It is not known why this occurs. However, it is known that the error occurs at conception and is not related to anything the mother did during pregnancy. It has been known for some time that the incidence of Down syndrome increases with advancing maternal age. However, 80% of children with Down syndrome are born to women under 35 years of age.

HEALTH ISSUES

Many children with Down syndrome have health complications beyond the usual childhood illnesses. Approximately 40% of the children have congenital heart defects. It is very important that an echocardiogram be performed on all newborns with Down syndrome in order to identify any serious cardiac problems that might be present.

Some of the heart conditions require surgery while others only require careful monitoring. Children with Down syndrome have a higher incidence of infection, respiratory, vision and hearing problems as well as thyroid and other medical conditions. However, with appropriate medical care most children and adults with Down syndrome can lead healthy lives. The average life expectancy of individuals with Down syndrome is 55 years, with many living into their sixties and seventies.

LEARNING & DEVELOPMENT

It is important to remember that while children and adults with Down syndrome experience developmental delays, they also have many talents and gifts and should be given the opportunity and encouragement to develop them.

TYPES OF DOWN SYNDROME

There are 3 chromosomal patterns that result in Down syndrome.

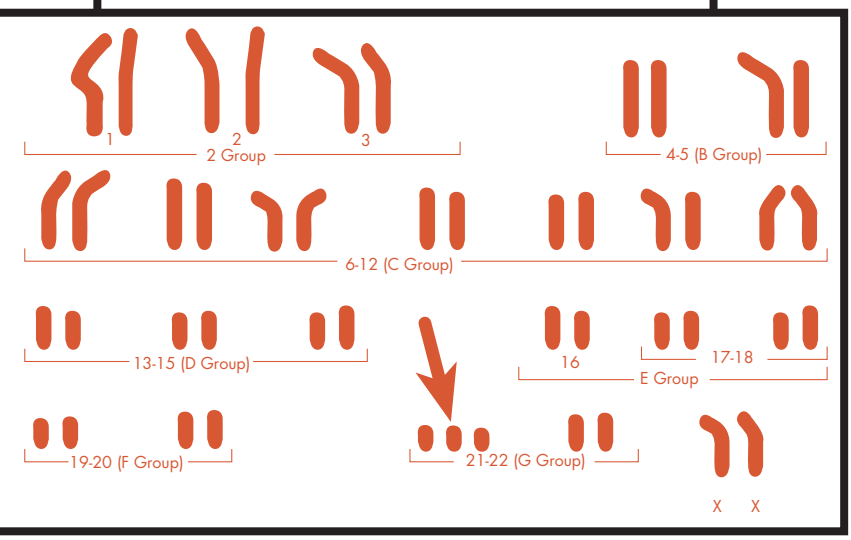
1. Trisomy 21 (nondisjunction) is caused by a faulty cell division that results in the baby having three #21 chromosomes instead of two. Prior to or at conception, a pair of #21 chromosomes in either the egg or the sperm fails to separate properly. The extra chromosome is replicated in every cell of the body. Ninety five percent of all people with Down syndrome have Trisomy 21.

2. Translocation accounts for only 3% to 4% of all cases. In translocation a part of chromosome #21 breaks off during cell division and attaches to another chromosome. The presence of an extra piece of the 21st chromosome causes the characteristics of Down syndrome. Unlike Trisomy 21, which is the result of random error in the early cell division, translocation may indicate that one of the parents is carrying chromosomal material that is arranged in an unusual manner.

Genetic counseling can be sought to ascertain more information when these circumstances occur.

3. Mosaicism occurs when nondisjunction of chromosome #21 takes place in one of the initial cell divisions after fertilization. When this happens, there is a mixture of two types of cells, some containing 46 chromosomes and some with 47. The cells with 47 chromosomes contain an extra 21st chromosome. Because of the "mosaic" pattern of the cells, the term mosaicism is used. This type of Down syndrome occurs in only one to two percent of all cases of Down syndrome.

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



Most children with Down syndrome have mild to moderate impairments but it is important to note that they are more like other children than they are different. Early Intervention services should be provided shortly after birth. These services should include physical, speech and developmental therapies. Most children attend their neighborhood schools, some in regular classes and others in special education classes. Some children have more significant needs and require a more specialized program.

Some high school graduates with Down syndrome participate in post-secondary education. Many adults with Down syndrome are capable of working in the community, but some require a more structured environment.

PRENATAL DIAGNOSIS

Two types of procedures are available to pregnant women: screening tests and diagnostic tests. The screening tests estimate the risk of the baby having Down syndrome. Diagnostic tests tell whether or not the baby actually has Down syndrome.

Screening Tests

- At this time the most commonly used screening test is "The Triple Screen." This is a combination of three tests that measure quantities of various substances in the blood. These tests are usually done between 15 and 20 weeks of gestation.
- Sonograms (ultrasounds) are usually performed in conjunction with other screenings. These can show some physical traits that are helpful in calculating the risk of Down syndrome.
- Screening tests do not accurately confirm the diagnosis of Down syndrome. In fact, false positives and false negatives frequently occur.

Diagnostic Tests

Three diagnostic tests are currently available:

- Amniocentesis is performed between 12 and 20 weeks gestation.
- Chorionic Villus Sampling (CVS) is conducted between 8 and 12 weeks.
- Percutaneous Umbilical Blood Sampling (PUBS) is performed after 20 weeks.