

Down Syndrome

Overview

Down syndrome is a genetic condition that occurs in one in every 733 births. It is the most frequently occurring chromosomal condition and is found in people of all races and economic levels. More than 400,000 people in the United States have Down syndrome.

A few of the common physical traits of Down syndrome are low muscle tone, small stature, an upward slant to the eyes, and a single deep crease across the center of the palm. Every person with Down syndrome is a unique individual and may possess these characteristics to different degrees or not at all.

People with Down syndrome have an increased risk for certain medical conditions such as congenital heart defects, respiratory and hearing problems, Alzheimer's disease, childhood leukemia, and thyroid conditions. However, many of these conditions are now treatable, so most people with Down syndrome lead healthy lives. Life expectancy for people with Down syndrome has increased dramatically in recent decades - from 25 in 1983 to 60 today.

People with Down syndrome experience cognitive delays, but the effect is usually mild to moderate and is not indicative of the many strengths and talents that each individual possesses. Children with Down Syndrome learn to sit, walk, talk, play, and do most other activities only somewhat later than their peers without Down syndrome.

Quality educational programs, a stimulating home environment, good health care, and positive support from family, friends and the community enable people with Down syndrome to develop their full potential and lead fulfilling lives. People with Down syndrome attend school and work, and participate in decisions that concern them, and contribute to society in many wonderful ways.

What is Down syndrome?

Down syndrome is the most common genetic condition. On in every 733 babies is born with Down syndrome. The most common form of Down syndrome is called Trisomy 21 because it involves an extra copy of the 21st chromosome.

What impact does Down syndrome have on society?

Individuals with Down syndrome are becoming increasingly integrated into society and community organizations, such as school, health care systems, work forces, and social and recreational activities. Individuals with Down syndrome possess varying degrees of intellectual disabilities, from very mild to severe. Most people with Down syndrome have IQs in the mild to moderate range of intellectual disability.

Due to advances in medical technology, individuals with Down syndrome are living longer than ever before. In 1910, children with Down syndrome were expected to survive to age nine. With the discovery of antibiotics, the average survival age increased to 19 or 20. Now, with recent advancements in clinical treatment, most particularly corrective heart surgeries, as many as 80% of adults with Down syndrome reach age 60, and many live even longer.

In the United States, approximately 400,000 families have a child with Down syndrome, and about 5,000 babies with Down syndrome are born each year. More and more Americans will interact with individuals with this genetic condition. Increasing the need for widespread public education and acceptance.

What is the cause of Down syndrome?

The additional copy of the 21st chromosome which causes Down syndrome can originate from either the father or the mother. Approximately 5% of the cases have been traced to the father.

Who has the highest risk of having a child with Down syndrome?

Down syndrome can occur in people of all races and economic levels. Older women have an increased chance of having a child with Down syndrome. A 35-year-old woman has about a one in 350 chance of conceiving a child with Down syndrome, and this chance increases gradually to one in 100 by age 40. At age 45 the incidence becomes approximately one in 30.

Since many couples are postponing parenting until later in life, the incidence of Down syndrome conceptions is expected to increase. Therefore, genetic counseling for parents is becoming increasingly important. Still many physicians are not fully informed about advising their patients about the incidences of Down syndrome, advancements in diagnosis, and the protocols for care and treatment of babies born with Down syndrome.

Why are medical researchers today so keenly interested in Down syndrome?

Down syndrome is a developmental condition. As researchers learn more about the molecular genetics and other aspects of Down syndrome, they also obtain valuable information about human development and can advance the study of many biological processes.

In addition, individuals with Down syndrome have a higher incidence of certain medical conditions, and the study of Down syndrome may yield important breakthroughs in those areas. Research in Down syndrome provides a way for looking at many important problems:

Heart disease: Up to 50% of individuals with Down syndrome are born *with* congenital heart conditions. The majority of heart conditions in children with Down syndrome can now be surgically corrected with resulting long-term health improvements. However, scientists continue to search for the cause of these heart conditions and look for means of prevention.

Alzheimer's disease: Estimates vary, but it is reasonable to conclude that 25% or more of individuals with Down syndrome over the age of 35 will develop the clinical signs and symptoms of Alzheimer's-type dementia.

Leukemia: Approximately one in every 100 individuals *with* Down syndrome will develop leukemia; or, to put it another way, 99% of people with Down syndrome will not develop leukemia. The majority of cases are categorized as acute megakaryoblastic leukemia, which tends to occur in the first three years of life, and for which there is a high cure rate. A transient form of leukemia is also seen in newborns with Down syndrome, disappearing spontaneously during the first two to three months of life.

Information provided by the National Down syndrome Society