Questions and Answers About Down Syndrome

Why should we be concerned about Down syndrome?

Chromosomal abnormalities are a widespread medical problem, with Down syndrome being the most common genetic condition. One in every 700 to 800 children is born with Down syndrome.

More than 50 percent of miscarriages are caused by a chromosomal abnormality. As many as 25 percent of all miscarriages are caused by a trisomy, which is the presence of three copies of a particular chromosome, rather than the normal two.

The most common form of Down syndrome is called Trisomy 21, because it involves an extra copy of the 21st chromosome. Twenty to 25 percent of children conceived with Down syndrome survive past birth.

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 mental retardation, from very mild to severe. Most people with Down syndrome have IQs in the mild to moderate range of mental retardation.

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, as many as 80 percent of adults with Down syndrome reach age 55, and many live even longer.

In the United States, approximately 350,000 families are affected by Down syndrome. Approximately 5,000 children with Down syndrome are born each year. As the mortality rate associated with Down syndrome is decreasing, the prevalence of individuals with Down syndrome in our society will increase. Some experts project that the number of people with Down syndrome will double in the next 10 years. More and more Americans will interact with individuals with this genetic condition, increasing the need for widespread public education and acceptance.

Is Down syndrome transmitted from the mother or father?

The additional genetic material which causes Down syndrome can originate from either the father or the mother. Approximately five percent of the cases have been traced to the father.

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

Down syndrome affects people of all races and economic levels. Women age 35 and older have a significantly increased risk of having a child with Down syndrome. A 35-year-old woman has a one in 400 chance of conceiving a child with Down syndrome and this chance increases gradually to one in 110 by age 40. At age 45 the incidence becomes approximately one in 35.

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 incidence of Down syndrome, advancements in diagnosis and the protocols for care and treatment of babies born with Down syndrome.

Why is it important to raise children with Down syndrome at home?

A greater understanding of Down syndrome and advancements in treatment of Down syndrome-related health problems have allowed people with Down syndrome to enjoy fuller and more active lives.

Children raised at home and included in all aspects of community life can best reach their potential and function in society with a greater degree of independence. Parental love, nurturing and support, as well as early intervention programs, educational opportunities

and community involvement, have a direct relationship to the degree that a person with Down syndrome is able to achieve his/her potential.

Why are medical researchers following Down syndrome work so closely?

Down syndrome is a developmental disorder. 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 problems 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 percent of individuals with Down syndrome are born with congenital heart defects. The majority of heart defects 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 this problem and look for means of prevention.

Alzheimer's disease: Estimates vary, but it is reasonable to conclude that 25 percent 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: Individuals with Down syndrome have a 15 to 20 times greater risk of developing 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.

Why hasn't Down syndrome received much attention in the past?

Even though Dr. Jerome Lejeune discovered in 1959 that it was an extra 21st chromosome that caused Down syndrome, it is only in the last few years that a focus has been placed on the study of the 21st chromosome.

In May 2000, researchers completed sequencing the approximately 225 genes on the 21st chromosome - only the second chromosome to be fully sequenced at this time. Although these findings will not have an immediate impact on the Down syndrome community, they will open the door to valuable research on this small set of genes.

Researchers continue to look for the genes related to the development of intelligence and the physical characteristics associated with Down syndrome. Once identified, it is hoped that the biochemical process which causes Down syndrome can be decoded, leading to the development of an intervention and cure. See the Research section of this Web site for more information about sequencing the 21st chromosome.

What is the National Down Syndrome Society doing to further research?

NDSS is currently sponsoring three scientists through the NDSS Science Scholar Award Program initiated in 1983. Under this grant program, each researcher receives $35,000 a year for two years to carry out scientific research into the causes and/or amelioration of Down syndrome. To date, more than 20 scholars have been supported by this program.

NDSS also sponsors a series of annual symposia focusing on Down syndrome research. This sharing of information, technology and the collective knowledge of the world's foremost researchers is expected to produce an acceleration of advancements in a variety of areas. The proceedings of these scientific symposia are published annually and are available through NDSS.