Fact Sheet: Down Syndrome

Down Syndrome is a chromosomal disorder that results in mild to severe learning disabilities and physical symptoms that include a small skull, extra folds of skin under the eyes, and a flattened nose bridge. Muscle tone throughout the body is usually low. The condition was formerly known as “mongolism” because the features of people with Down syndrome were thought to resemble those of Mongolian Asians. **This term is now considered offensive and inappropriate and is no longer used.**

Down syndrome occurs in about 1 out of every 800 births worldwide.

In the United States each year, about 1,600 babies are born with this condition. Down syndrome results when a person inherits all or part of an extra copy of chromosome 21. This can occur in a variety of ways, the causes of which are unknown. The most common chromosomal abnormality that produces Down syndrome (accounting for about 95 percent of all cases) is Trisomy 21, a defect in which an extra, third copy of chromosome 21 is present in every cell in the body. The risk of Trisomy 21 is directly related to the age of the mother. The number of Down syndrome births is relatively low for 18-year-old mothers—about 1 in 2,100 births. In the later childbearing years the risk increases significantly—from 1 in 1,000 births for 30-year-old women to 1 in 100 births for 40-year-old women.

Two other chromosomal abnormalities cause Down syndrome and occur in about 2 to 3 percent of all cases. The first, translocation, takes place when a child inherits a small, extra piece of the 21st chromosome that is attached to another chromosome. If, in addition to the translocation, two normal 21st chromosomes are also present, the person will have some of the features of Down syndrome. If there is only one normal 21st chromosome, the person will not display symptoms but the children may inherit Down syndrome. Mosaic Down syndrome results from a second type of chromosomal abnormality in which only some cells in the body have an extra chromosome.

There is no cure for Down syndrome. However, prenatal tests are available to identify fetuses with the disorder. The American College of Obstetricians and Gynecologists recommends that the so-called triple-screen blood test be offered to all pregnant women. This test measures the levels of three chemicals in the blood of the pregnant woman to indicate the baby's risk of Down syndrome. If the risk is high, amniocentesis, a procedure for removing a sample of the amniotic fluid surrounding the fetus, is administered to confirm the findings from the blood tests. Fetal cells are present in the amniotic fluid and can be checked for the presence of the chromosomal disorder.

People with Down syndrome are subject to a variety of medical conditions. Heart abnormalities that may require surgery are present in about half of all Down syndrome cases. Thyroid problems (underproduction or overproduction of thyroid hormones) affect 10 to 20 percent of people with Down syndrome, but these problems respond well to treatment. The risk of acute leukemia is somewhat increased, although treatment is successful in the majority of cases.

There have been dramatic increases in the survival rates of people with Down syndrome since the 1970s. As the risks of medical problems specific to Down syndrome have become known, doctors are now able to recognize those problems earlier, and develop more effective treatments. Today, 44 percent of people with Down syndrome survive to age 60, and this life expectancy is slowly approaching that of people without Down syndrome.

Although people with Down syndrome have a range of learning disabilities, physicians, educators, and parents now recognize that these people's achievements may be most influenced by what is expected of them. This so-called environmental expectation is perhaps the most important factor in determining the educational and vocational potential of people with Down syndrome. On the
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other hand, intelligence-quotient test scores, once considered an authoritative indicator of educational potential, are now seen to be of questionable value.

Educational and vocational opportunities have also advanced. In the recent past, children with Down syndrome were relegated to institutions, receiving minimal social interaction or educational opportunities. Today, children with Down syndrome usually remain with their families and are enrolled in public schools. Often they attend regular classes and learn skills such as reading and writing alongside children without Down syndrome. Adults with Down syndrome are employed in a range of fields. Some may live in supervised group homes, while others live independently.

Information from: Encyclopedia Article from Encarta Contributed By: Phil Mattheis, B.S., M.D.