Down Syndrome			
Staff Name		Date	
Read the information about this topic, print the questions at the end of the reading material, and then answer the questions. When you have completed answering the questions, compare your answers with the answers provided at the end of this document. Return your completed questions to the office.			

DEFINITION

Down syndrome is set of mental and physical symptoms that result from having an extra copy of Chromosome 21. Down syndrome is the most common and readily identifiable chromosomal condition associated with mild to moderate mental retardation and associated medical problems. It is caused by a chromosomal abnormality. For an unexplained reason, an accident in cell development results in 47 instead of the usual 46 chromosomes. This extra chromosome changes the orderly development of the body and brain. To confirm the diagnosis, a blood test called a *chromosomal karyotype* is done to determine if extra material from chromosome 21 is present.

A newborn baby with Down syndrome often has recognizable physical features, including a flat facial profile, an upward slant to the eye, a short neck, abnormally shaped ears, white spots on the iris of the eye (called Brushfield spots), and a single, deep transverse crease on the palm of the hand. However, a child with Down syndrome may not possess all of these features; some of these features can even be found in the general population. Caring for a child with Down syndrome frequently requires much time and energy and parents of newborn children with Down syndrome should seek the advice of a knowledgeable pediatrician and the many Down syndrome support groups and organizations available.

For centuries, people with Down syndrome have been alluded to in art, literature, and science. In the 19th century John Langdon Down, an English physician, published an accurate description of a person with Down syndrome. It was this work, published in 1866, which earned John Down the recognition as the "father" of the syndrome. Throughout the 20th century, advances in medicine and science enabled researchers to investigate the characteristics of people with Down syndrome. In 1959, the French physician, Jerome Lejeune, identified Down syndrome as a chromosomal anomaly when he observed 47 chromosomes present in each cell of individuals with Down syndrome instead of the usual 46. It was later determined that an extra partial or complete 21st chromosome results in the characteristics associated with Down syndrome.

Researchers have established that the likelihood that a reproductive cell will contain an extra copy of chromosome 21 increases dramatically as a woman ages. The chance of having a baby with Down syndrome increases as a woman gets older—from about 1 in 1,250 for a woman who gets pregnant at age 25, to about 1 in 100 for a woman who gets pregnant at age 40. Therefore, an older mother is more likely than a younger mother to have a baby with Down syndrome, but older mothers account for only about 9% of all live births each year and 25% of Down syndrome births. In other words, most babies with Down syndrome are born to women under age 35 because more younger women have babies.

Because the chances of having a baby with Down syndrome increase with the age of the mother, many health care providers recommend that women over age 35 have prenatal testing for the condition to enable parents to prepared for the baby's special needs.

Parents who already have a baby with Down syndrome or who have abnormalities in their own chromosome 21 are also at higher risk for having a baby with Down Syndrome.

Many people with Down syndrome have IQs that fall in the mild to moderate range of retardation. Some are so mildly affected that they live independently and are gainfully employed. One of the most frequently occurring chromosomal abnormalities, Down syndrome occurs in one out of 800 live births, in all races and economic groups. Approximately 4,000 children with Down syndrome are born in the U.S. each year and more than 350,000 people in the U.S. have this genetic condition.

There is no way to know the intellectual or physical capabilities of a Down child, or any other child. Children and adults with Down syndrome have a wide range of abilities and talents. A person with Down syndrome may be very healthy or they may present unusual and demanding medical and social problems at virtually every stage of life. However, every person with Down syndrome is a unique individual, and their disabilities range from severe to minimal.

CHARACTERISTICS

Most people with Down syndrome fall in the mild to moderate range of mental retardation which limits intellectual abilities and adaptive behaviors (conceptual, social, and practical skills people use to function in everyday lives). They may have delayed language development and slow motor development. There are over 50 clinical signs of Down syndrome, but it is rare to find all or even most of them in one person. Some common physical characteristics of Down syndrome include:

– Small stature
– Poor muscle tone, loose ligaments
– Hyperflexibility (excessive ability to extend the joints)
– Short, broad hands with a single, deep crease across the palm on one or both hands
– Small, broad feet and hands with short toes and fingers
– Flat face with upward slant to the eye with folds of skin at the inner corners (epicanthal fold), short neck, short, low-set ears

– Small oral cavity

CAUSES

The human body is made of cells. All cells contain a center, called a nucleus, in which genes are stored. Genes, which carry the codes responsible for all our inherited characteristics, 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. When the reproductive cells, the sperm and ovum, combine at fertilization, the fertilized egg that results contains 23 chromosome pairs. A fertilized egg that will develop into a female contains chromosome pairs 1 through 22, and the XX pair. A fertilized egg that will develop into a male contains chromosome pairs 1 through 22, and the XY pair. Down syndrome occurs when some or all of a person's cells have an extra full or partial copy of chromosome 21 (when the fertilized egg contains **extra material** from chromosome number 21).

The Genetic Variations That Can Cause down Syndrome
Three genetic variations can cause Down syndrome:
– Trisomy 21
– Mosaicism
– Translocation

Trisomy 21
In most cases, approximately 92-95% of the time, Down syndrome is caused by the presence of an **extra chromosome 21 in all cells** of the individual. In such cases, the extra chromosome originates in the development of either the egg or the sperm. Consequently, when the egg and sperm unite to form the fertilized egg, three--rather than two--chromosomes 21 are present. As the embryo develops, the extra chromosome is repeated in every cell. This condition, in which three copies of chromosome 21 are present in all cells of the individual, is called trisomy 21.

Mosaicism (Mosaic Trisomy 21)
The least common form of Down syndrome (approximately 2-4% of cases) is due to mosaic trisomy 21. This situation is similar to simple trisomy 21, but, in this instance, the **extra chromosome 21** is present **in some cells** of the individual, but not all. For example, the fertilized egg may have the right number of chromosomes, but, due to an error in chromosome division early in embryonic development, some cells acquire an extra chromosome 21. Thus, an individual with Down syndrome due to mosaic trisomy 21 will typically have 46 chromosomes in some cells, but will have 47 chromosomes (including an extra chromosome 21) in others. In this situation, the range of the physical problems may vary, depending on the proportion of cells that carry the additional chromosome 21.

Translocation (Translocation Trisomy 21)
In trisomy 21 and mosaic trisomy 21, Down syndrome occurs because some or all of the cells have 47 chromosomes, including three chromosomes 21. However, approximately 3-4% of individuals with Down syndrome have cells containing 46 chromosomes, but still have the features associated with Down syndrome. In such cases, **material from one chromosome 21 gets stuck or translocated onto another chromosome**, either prior to or at conception. In such situations, cells from individuals with Down syndrome have two normal chromosomes 21, but also have additional chromosome 21 material on the translocated chromosome. Thus, there is still too much material from chromosome 21, resulting in the features associated with Down syndrome.

The cause of the extra full or partial chromosome is still unknown. It is not caused by environmental factors or anything the mother does before or during her pregnancy. Maternal age is the only factor that has been linked to an increased chance of having a baby with Down syndrome resulting from trisomy 21 or mosaicism.

Maternal age, however, is not linked to the chance of having a baby with translocation. Most cases are sporadic, chance events, but in about one third of translocation cases, one parent is a carrier of a translocated chromosome. For this reason, the chance of translocation in a second pregnancy is higher than that seen in trisomy 21.

Most of the time, the occurrence of Down syndrome is due to a random event that occurred during formation of the reproductive cells, the ovum or sperm. Down syndrome is not attributable to any behavioral activity of the parents or environmental factors. The probability that another child with Down syndrome will be born in a subsequent pregnancy is about 1 in 100, although age may also be a factor.

HEALTH ISSUES/COMPLICATIONS

Besides having a distinct physical appearance, children with Down syndrome frequently have specific health-related problems. A variety of health conditions may often be seen in people who have Down syndrome with varying incidence and severity, including:
• Congenital heart disease
  Approximately half of the children with Down syndrome have congenital heart disease.
• Respiratory diseases
  Including middle ear infections, tonsillitis, pneumonia and pulmonary hypertension (high blood pressure in the lungs often associated with congenital heart disease). Compared to the general population, individuals with Down syndrome have a 12% higher mortality rate from infectious diseases if these infections are left untreated and unmonitored.
• Intestinal problems
  Including blocked small bowel or esophagus and Celiac disease which is a digestive disease that damages the small intestine and interferes with absorption of nutrients from food.
• Hearing and eye problems (such as cataracts, pterygium)
• Thyroid dysfunctions
• Skeletal problems
  Including susceptibility to transient myelodysplasia, or the defective development of the spinal cord and Atlantoaxial Instability which is a misalignment of the top two vertebrae of the neck makes these individuals more prone to injury if they participate in activities which overextend or flex the neck. Although this misalignment is a potentially serious condition, proper diagnosis and restriction from sports and activities which place stress on the neck can help prevent serious injury.
• Dementia—similar to Alzheimer's
• Seizure disorders

Seizure disorders, though less prevalent than some of the other associated medical conditions, still affect between 5 and 13% of individuals with Down syndrome, a 10-fold greater incidence than in the general population.

• Leukemia
Individuals with Down syndrome are10-15 times more likely than other children to develop leukemia, a potentially fatal blood disease.

• Obesity
Children with Down syndrome may have a tendency to become obese as they grow older. Besides having negative social implications, this weight gain threatens these individuals' health and longevity. A supervised diet and exercise program may help reduce this problem.

• Developmental delays
Development of speech and language abilities may take longer than expected. Hypotonia, or poor muscle tone, may cause feeding difficulties resulting in nutritional deficiencies as well as delays in ability to turn over, sit, stand, and respond.

TREATMENT/SUPPORT

Down syndrome is a genetic condition, not a condition that can be cured. However, early intervention can help many people with Down syndrome live productive lives well into adulthood. 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. Today people with Down syndrome live at home with their families and are active participants in the educational, vocational, social and recreational activities of the community. They are integrated into the regular education system, and take part in sports, camping, music, art programs and all the other activities of their communities. In addition, they are socializing with people with and without disabilities, and as adults are obtaining employment and living in group homes and other independent housing arrangements, such as supported living.

In general, individuals with Down syndrome will benefit from:
- caregiver education
- early intervention
- monitoring health
- weight control
- therapy, including physical, speech, music, and occupational
- stimulating environment
- encouragement

Children with Down syndrome can often benefit from speech therapy, occupational therapy, and exercises for gross and fine motor skills. Special education and attention at school and inclusion into regular classes at school may also be of benefit.

Federal laws (Public Law 94-142) are in place to ensure each state has as a goal that "all handicapped children have available to them a free public education and related services designed to meet their unique needs." The decision of what type of school a child with Down syndrome should attend is an important one, made by the parents in consultation with health and education professionals. A parent must decide between enrolling the child in a school where most of the children do not have disabilities (inclusion) or sending the child to a school for children with special needs. Inclusion has become more common over the past decade.

Parents of other children with Down syndrome are often valuable sources of information and support. Parents should keep in mind that children with Down syndrome have a wide range of abilities and talents, and each child develops at his own particular pace. It may take children with Down syndrome longer than other children to reach develop mental milestones, but many of these milestones will eventually be met. Parents should make a concerted effort not to compare the developmental progress of a child with Down syndrome to the progress of other siblings or even to other children with Down syndrome.

Shortly after a diagnoses of Down syndrome is confirmed, parents should be encouraged to enroll their child in an infant development/early intervention program. These programs offer parents special instruction in teaching their child language, cognitive, self-help, and social skills, and specific exercises for gross and fine motor development. Early intervention, specialized programs and related resources, may include special educators, speech therapists, occupational therapists, and social workers. It is recommended that stimulation and encouragement be provided to children with Down syndrome. Research has shown that stimulation during early developmental stages improves the child's chances of developing to his fullest potential. Continuing education, positive public attitudes, and a stimulating home environment have also been found to promote the child's overall development.

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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 potential.

Just as in the normal population, there is a wide variation in mental abilities, behavior, and developmental progress in individuals with Down syndrome. Because of the range of ability in children with Down syndrome it is important for families and educators to place few limitations on potential capabilities. In addition, effective training includes:
– emphasizing concrete concepts rather than abstract ideas
– teaching tasks in a step-by-step manner
– providing frequent reinforcement
– using consistent feedback
– teaching consistent routines

Improved public acceptance and increased opportunities for adults with disabilities to live and work independently in the community have expanded goals for individuals with Down syndrome. Independent living centers, group shared and supervised apartments, and support services in the community are important resources for persons with disabilities.

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. Home-based care and community living give them the opportunity to socialize and benefit from such interactions.

The Americans with Disabilities Act (ADA) makes it illegal for an employer of more than 15 individuals to discriminate against qualified individuals in application procedures, hiring, advancement, discharge, compensation, job training, and other terms of employment. The ADA requires that an employer provide reasonable accommodation for individuals who are qualified for a position. More information about the ADA can be obtained from the Office of Civil Rights of the U.S. Department of Health and Human Services, Washington, DC, 20201.

Like all teenagers, individuals with Down syndrome undergo hormonal changes during adolescence. Therefore, teenagers with Down syndrome should be educated about their sexual drives. Scientists have medical evidence that males with Down syndrome generally have a reduced sperm count and rarely father children. Females with Down syndrome have regular menstrual periods and are capable of becoming pregnant and carrying a baby to term. Many people with Down syndrome form meaningful relationships and eventually marry.

The life expectancy for people with Down syndrome has increased substantially. In 1929, the average life span of a person with Down syndrome was nine years. 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. Today, it is common for a person with Down syndrome to live to age fifty and beyond. In addition to living longer, people with Down syndrome are now living fuller, richer lives than ever before as family members and contributors to their community. Now that people with Down syndrome are living longer, the needs of adults with Down syndrome are receiving greater attention. With assistance from family and caretakers, many adults with Down syndrome have developed the skills required to hold jobs and to live semi-independently.

Premature aging is a characteristic of adults with Down syndrome. In addition, dementia, or memory loss and impaired judgment similar to that occurring in Alzheimer disease patients, may appear in adults with Down syndrome. This condition often occurs when the person is younger than forty years old. Family members and caretakers of an adult with Down syndrome must be prepared to intervene if the individual begins to lose the skills required for independent living.

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.

LONG TERM OUTLOOK

Today, individuals with Down syndrome are active participants in the educational, vocational, social and recreational aspects of our communities. In fact, there are more opportunities than ever before for individuals with Down syndrome to develop their abilities, discover their talents and realize their dreams. For example, more teens and adults with Down syndrome each year are graduating from high school, going to college, finding employment and living independently.

The opportunities currently available to individuals with Down syndrome have never been greater. However, it is only through the collective efforts of parents, professionals, and concerned citizens that acceptance is becoming even more widespread. It is the mission of the National Down Syndrome Society to ensure that all people with Down syndrome are provided the opportunity to achieve their full potential in all aspects of their lives.

In almost every community of the United States there are parent support groups and other community organizations directly involved in providing services to families of individuals with Down syndrome.

Children with Down syndrome have been included in regular academic classrooms in schools across the country. In some instances they are integrated into specific courses, while in other situations students are fully included in the regular classroom for all subjects. The degree of mainstreaming is based in the abilities of the individual; but the trend is for full inclusion in the social and educational life of the community.

Businesses are seeking young adults with Down syndrome for a variety of positions. They are being employed in small and medium sized offices: by banks, corporations, nursing homes, hotels and restaurants. They work in the music and entertainment industry, in clerical positions and in the computer industry. People with Down syndrome bring to their jobs enthusiasm, reliability, and dedication.

Recently, it has been suggested that children with Down syndrome might benefit from medical intervention that includes amino acid supplements and a drug known as Piracetam. Piracetam is a psychoactive drug that some believe may improve cognitive function. However, there have been no controlled clinical studies conducted to date using Piracetam to treat Down syndrome in the U.S. or elsewhere that show its safety and efficacy.

Researchers have identified the genes that cause the characteristics of Down syndrome and are working to develop models to analyze the developmental consequences of Down syndrome. Studying these models at varying stages of development will enhance our basic understanding of Down syndrome and facilitate the development of effective interventions and treatment strategies.

Additional Resources for Down Syndrome

Administration on Developmental Disabilities
Administration for Children and Families
U.S. Department of Health and Human Services
Mail Stop: HHH 300F
370 L'Enfant Promenade S.W.
Washington, DC 20447
(202) 690-6590
http://www.acf.dhhs.gov/programs/add/

American Speech, Language and Hearing Association
10801 Rockville Pike
Rockville, MD 20852
1-800-638-8255 or 1-888-321-ASHA
http://www.asha.org/

Learning Disabilities Association of America
4156 Library Road
Pittsburgh, PA 15234-1349
(412) 341-1515 or 1-888-300-6710
http://www.ldanatl.org/

March of Dimes
1275 Mamaroneck Avenue
White Plains, NY 10605
(914) 428-7100
1-888-MODIMES (1-888-663-4637)
http://www.modimes.org/

National Down Syndrome Congress
1370 Center Drive, Suite 102
Atlanta, GA 30338
1-800-232-6372
(770) 604-9500
http://www.ndsccenter.org/

National Down Syndrome Society
666 Broadway
New York, NY 10012
1-800-221-4602
(212) 460-9330
http://www.ndss.org/

National Information Center for Children and Youth with Disabilities
P.O. Box 1492
Washington, DC 20013-1492
1-800-695-0285
(202) 884-8200
http://www.nichcy.org/

Mid-Atlantic Regional Human Genetics Network (MARHGN)
(genetic counseling)
Curtis Coughlin II, MS, MARHGN Coordinator
MARGHN c/o Christiana Health Care Services Genetics Room 1988
4755 Ogletown-Stanton Road
P.O. Box 6001
Newark, DE 19718
(302) 733-6732
http://www.pitt.edu/~marhgn/

National Society of Genetic Counselors
233 Canterbury Drive
Wallingford, PA 19086-6617
(610) 872-7608
http://www.nsgc.org/

The Arc of the United States
1010 Wayne Avenue, Suite 650
Silver Spring, MD 20910
(301) 565-3842
(301) 565-3843 - Fax
http://www.thearc.org

REFERENCES

http://www.ndss.org/index.php?option=com_content&task=view&id=1812&Itemid=95

http://www.nichd.nih.gov/health/topics/Down_Syndrome.cfm

http://www.nichd.nih.gov/publications/pubs/downsyndrome.cfm

http://www.nichcy.org/pubs/factsheet/fs4txt.htm