Guide for New and Expectant Parents

Table of Contents – Section 2

Facts about Down Syndrome – Section 2
Down Syndrome - What a New or Expectant Parent Should Know.............. 2-1
A Promising Future........................................................................................................ 2-6
Facts about Down Syndrome...................................................................................... 2-8
Myths and Truths........................................................................................................... 2-9

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Visit any library and you will most likely find an array of books regarding Down syndrome. Unfortunately, many of those texts are outdated and do not provide accurate information on the subject. There has been a great deal of research done on Down syndrome in recent years, and new discoveries are continually being made. Society has also taken new approaches toward persons with Down syndrome. The available medical treatments, educational opportunities, living arrangements and employment prospects are improving each year.

With such a wide range of accessible information on Down syndrome, new parents may be overwhelmed and need assistance in discerning the most pertinent facts. The most common issues that envelop new parents are:

- What is Down Syndrome? (discussed below)
- Early Development and Intervention (see Section 3)
- Health & Medical Concerns (see Section 4)

**What is Down Syndrome?**

Down syndrome is named after John Langdon Down, an English physician who first described the physical characteristics of the syndrome in 1866. It is one of the most frequently occurring chromosomal abnormalities, affecting approximately one in 691 live births. Down syndrome is caused by the presence of an extra 21st chromosome, in addition to the normal pair of 21st chromosomes, in each of a person's cells. Dr. Jerome LeJeune, a French biologist, discovered this extra chromosome in 1959.

Normally, each of a person's cells has 46 chromosomes – a pair each of chromosomes 1-22 and a pair of sex chromosomes. The extra 21st chromosome results in a total of 47 chromosomes in the cells of persons with Down syndrome. At conception, the sperm and the egg each provide half of a chromosomal set to the fertilized cell. If a pair of chromosomes fails to split during the cell divisions that lead to the development of the egg or the sperm, it leaves one or the other with two chromosomes of the same kind. When this pair matches up with a third chromosome during fertilization, this results in a trisomy and a cell with a total of 47 chromosomes (Siwolop and Mohs, 1985). This is called *Trisomy 21.*
Any chromosomes can have a trisomy, but most trisomies result in miscarriages. Because the 21st chromosome is small, the embryo can often survive with the extra genetic material of a third chromosome and become a child with Down syndrome.

Down syndrome also encompasses two other genetic conditions: mosaicism and translocation. Mosaicism occurs when nondisjunction (failure of chromosome pairs to properly separate, as above) of chromosome 21 takes place in one of the initial cell divisions after fertilization, causing a person to have 46 chromosomes in some of their cells and 47 in others. The least common form of Down syndrome, mosaicism accounts for only one to two percent of all cases.

Translocation, which accounts for three to four percent of cases of Down syndrome, occurs when part of chromosome 21 breaks off during cell division and attaches to another chromosome, usually chromosome 14. While the total number of chromosomes in the cells remains 46, the presence of an extra part of chromosome 21 causes the characteristics of Down syndrome.

The cause of the extra full or partial chromosome is still unknown, but we do know that 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 nondisjunction. A 35-year-old woman has a one in 350 chance of conceiving a child with Down syndrome. By age 45, the incidence has increased to one in 30. However, because younger women have higher fertility rates, 80 percent of babies with Down syndrome are born to women under the age of 35. Once a woman has given birth to a baby with Down syndrome, the chance of having a second child with Down syndrome is about one in 100, although age may also be a factor.

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 nondisjunction.

Regardless of the type of Down syndrome a child has, the most important thing to know is that nothing the parents did or didn’t do caused it.

Prenatal Testing
There are two types of tests for Down syndrome that can be performed before your baby is born: screening and diagnostic tests. Prenatal screenings estimate the chance of the fetus having Down syndrome. These tests do not tell you for sure whether your baby has Down syndrome; they only provide a risk assessment. Diagnostic tests, on the other hand, can provide a definitive diagnosis with almost 100 percent accuracy.

There are two types of prenatal screening tests available: maternal serum screening and ultrasound (sonogram) screening. Maternal serum screening tests measure quantities of various substances in the blood of the mother, including alpha-fetoprotein and the hormones estriol and human chorionic gonadotropin. Together with a woman’s age, these are used to estimate her chance of having a child with Down syndrome. Typically offered between 15 and 20 weeks of gestation, maternal serum screening tests are only
able to accurately detect about 60 percent of fetuses with Down syndrome. Many women who undergo these tests will be given false-positive readings, and some will be given false-negative readings.

Because maternal serum screening tests are of limited value, they are often performed in conjunction with a detailed sonogram to check for “markers” (characteristics that some researchers feel may have a significant association with Down syndrome). Recently, researchers have developed a maternal serum/ultrasound/age combination that can yield a much higher accuracy rate at an earlier stage in the pregnancy. Researchers are also developing a definitive maternal blood test, to be offered in the first trimester of pregnancy, which will pose no risk to the fetus.

Prenatal screening tests are routinely offered to all pregnant women on the recommendation of the American College of Obstetrics and Gynecology (ACOG); however, pregnant women can choose not to have it done. If the estimate determined by prenatal screening is high, doctors will often advise a mother to undergo diagnostic testing.

The diagnostic procedures available for prenatal diagnosis of Down syndrome are chorionic villus sampling (CVS), amniocentesis and percutaneous umbilical blood sampling (PUBS). These procedures, which carry a small risk of miscarriage, are about 98 to 99 percent accurate in the detection of Down syndrome. Amniocentesis is usually performed between 15 and 22 weeks of gestation, CVS between nine and 14 weeks, and PUBS after 18 weeks.

If you have any questions about these procedures, do not hesitate to ask your doctor. It is important that you receive accurate information and understand all your options. Whether or not to undergo a prenatal screening or diagnostic test is a personal decision, and expectant parents must make the choice that is best for them.

**Diagnosis in a Newborn**

Even though there are many prenatal tests available for Down syndrome, most cases of Down syndrome are diagnosed after the baby is born. Doctors will usually suspect Down syndrome if certain physical characteristics are present. Some of the traits common to babies with Down syndrome include:

- flattened profile with a small nose and depressed nasal bridge
- flattened back of the head
- upward slanting eyes with folds of skin at the inner corners (epicanthic folds)
- smaller ears that are lower set
- low muscle tone and excessive joint flexibility
- a single horizontal crease (simian crease) across the palm of the hand
- a large tongue in relation to the size of the mouth
- more space between the large and second toes
Not all babies with Down syndrome have all these characteristics, and many of these features can be found to some extent in individuals who do not have the condition. Therefore, doctors must perform a special test called a karyotype before making a definitive diagnosis.

To obtain a karyotype, doctors draw a blood sample to examine your baby’s cells. They use special tools to photograph the chromosomes and then group them by size, number and shape. By examining the karyotype, doctors can tell definitively whether or not your baby has Down syndrome.

**Your Baby’s Development**

An additional chromosome means that there is excess genetic material in your baby’s cells. While this will affect your child’s development, it is important to realize that it is not a blueprint that determines his or her potential. Down syndrome is a condition your child has – it’s not who your child is. As is true for all people, the skills and knowledge he or she acquires will be a unique combination of innate abilities and life experiences.

In most ways, your baby will be just like other infants. Every baby needs to be fed, held and most of all, loved. But there are certain health and developmental concerns commonly associated with Down syndrome that you should to be aware of.

Individuals with Down syndrome are at an increased risk for certain health complications. Babies, in particular, are more likely to have heart problems, hearing loss and respiratory infections; however, advances in medicine have rendered the majority of these health problems treatable.

In addition, all people with Down syndrome experience delays in their cognitive and physical development. Cognitive delays are usually mild to moderate, and they are not indicative of the many strengths and talents that each individual possesses. Low muscle tone and other physical characteristics associated with Down syndrome can affect how soon your baby is able to sit up, walk and speak. Rest assured, though, that your child will learn how to do these and many other activities, only possibly somewhat later than his or her peers without Down syndrome.

Good medical care and early intervention can provide a strong foundation for your child’s optimal development. Sections 3 and 4 of this guide will help you begin learning about what you can do to help your baby get off to the best start possible!

In the past, it was incorrectly assumed that all people with Down syndrome had severe mental impairments. Current research indicates that the majority of people with Down syndrome have mild to moderate impairments. When a baby is born with Down syndrome, quite frequently Intelligence Quotient (IQ) becomes the main focal point. Many people attempt to predict an infant’s IQ or potential, and yet we know it is impossible to determine any infant’s IQ.

As with all children, youngsters with Down syndrome acquire new skills and develop individually as they grow, albeit at a slower pace for children with Down syndrome.
There are individual differences in the development of all children. **It is impossible to predict the future strengths and weaknesses of ANY baby.**

In general, the current achievement norm for teenagers and adults with Down syndrome is probably not a good indicator of what will be typical in the future. The common attitude toward all people with disabilities is more and more one of acceptance. Education, job and housing opportunities have increased significantly during the past years and, it appears, will continue to do so.

As you attempt to imagine your child’s future, it is helpful to realize that there is no standard profile of a person with Down syndrome. As is true for everyone, the skills and knowledge he or she acquires will depend on life experiences as well as innate abilities. Many parents find that their increased participation and the additional effort it takes a child with Down syndrome to accomplish things makes each newly acquired skill feel like a major success. A great deal of satisfaction can be derived from the most minor accomplishments. At the same time, it can also be frustrating when a child progresses slowly and efforts seem futile. There are special charts that provide some indication of average ages for reaching development milestones in a child with Down syndrome (see Section 3). Remember, these are only averages and each child differs, just as typically-developing children do.

There are many other issues regarding Down syndrome that will become concerns as children transition from school to adulthood. The article entitled, *A Promising Future*, which follows, discusses several of these concerns.

Portions of this article are from *A Promising Future Together: A Guide for New and Expectant Parents*, National Down Syndrome Society, 2005

Additionally, the following sources were referenced:
- National Down Syndrome Society, 666 Broadway, New York, NY 10012
- Siwolop, Sana and Mohs, Mayo, *“The War on Down Syndrome”, Discover, February 1985*
- Tingey, Carol, *Down Syndrome: A Resource Handbook*
A Promising Future

Your child has been born, or is about to be delivered, into a world that offers more opportunities than ever before for people with Down syndrome to reach their full potential. Every day, we are expanding our knowledge about how individuals with Down syndrome learn and the best ways to support their development. Scientific research is constantly yielding new information about the causes of Down syndrome and associated conditions. Also, many local and national advocacy organizations are working tirelessly to promote legislation that advances the rights of individuals with disabilities. All these efforts have opened up many doors for people with Down syndrome to pursue their dreams.

What does the future hold for people with Down syndrome?

There is still much progress to be made, but with the positive developments we’ve seen in recent years, you have every reason to be optimistic about your baby’s future. For example, today the majority of children with Down syndrome are included in regular education classrooms alongside their peers, and research has shown that this inclusion has positive effects on both the academic and social experiences of students with and without disabilities.

Many individuals with Down syndrome are also graduating from high school and going on to college. The Individuals with Disabilities Education Act requires that your child have a plan in place to ensure a successful transition to life after high school, and many new postsecondary programs especially for people with disabilities are available. In addition, we are seeing an increase in the amount of meaningful, satisfying employment opportunities available to people with Down syndrome and more options for independent living. Many people with Down syndrome have jobs and live on their own. Some even have the opportunity to get married.

The fact that these options are available today gives us reason to believe that for the next generation of people with Down syndrome – including your child – the future is even brighter.

How can I give my baby the best chance at a promising future?

No one can tell you your child’s potential, but there are many things you can do to give your child the best chance at a successful and happy life. First and foremost, your child will need lots of love, affection and support for healthy development. Just like all kids, children with Down syndrome will have their own unique talents and abilities, and it is important to recognize and celebrate those accomplishments. During the first few years of life, your child will likely receive many early intervention services, including physical, speech and language, and occupational therapy. Still, instead of emphasizing what your child can’t do, focus on what he or she can do! We are used to doing this with typically-developing children, and it should be no different for kids with disabilities.

Of course, babies and children with Down syndrome will have certain developmental challenges that will need special attention. In addition to providing your baby with varied experiences and constant opportunities for growing and learning, you’ll also want to
learn as much as you can about Down syndrome. There are many successful strategies for addressing specific challenges, but often it’s up to parents to track them down. Fortunately, there are many organizations and resources available to you that can provide information on specific topics.

Lastly, although your child with Down syndrome may need more attention from time to time, do your best to treat him or her the same as your other children and have similar expectations. Inclusion starts in the family. By going out into the community and enjoying all the same activities that other families enjoy, you will be teaching your child that he or she has the same right as everyone else to live a full and active life. You will also be showing others that people with Down syndrome are more like the rest of us than they are different.

As a parent, you are a natural advocate for your child. You will probably come across many people who do not know very much about Down syndrome, and you’ll be able to share information with them about your child’s abilities and the potential of people born with this condition. As you become more comfortable in your role, you may find that you want to advocate for people with Down syndrome in more formal ways, and there are many opportunities to get involved!

Facts About Down Syndrome

Presented by the National Down Syndrome Congress

- Down syndrome is a chromosome disorder that usually causes delays in physical, intellectual and language development.
- The exact causes of the chromosomal rearrangement and primary prevention of Down syndrome are currently unknown.
- Down syndrome is one of the leading clinical causes of mental impairment in the world – it is not related to race, nationality, religion or socioeconomic status.
- The incidence of Down syndrome in the United States is estimated to be one in every 691 live births.
- Of all children born in this country annually, approximately 6,000 will have Down syndrome.
- There are approximately 350,000 families in the United States affected by Down syndrome.
- Women under 35 years of age give birth to more than 80% of children with Down syndrome.
- There is wide variation in mental abilities, behavior and physical development in individuals with Down syndrome. Each individual has his or her own unique personality, capabilities and talents.
- Individuals with Down syndrome benefit from loving homes, early intervention, special education, appropriate medical care and positive public attitudes.
- In adulthood, many persons with Down syndrome hold jobs, live independently and enjoy recreational opportunities in their communities.
Down Syndrome: Myths and Truths

Myth: *Down syndrome is a rare genetic disorder.*

Truth: Down syndrome is the most commonly occurring genetic condition. One in every 691 live births is a child with Down syndrome, representing approximately 6,000 births per year in the United States alone. Today, more than 400,000 people in the United States have Down syndrome.

Myth: *People with Down syndrome are severely impaired.*

Truth: Most people with Down syndrome have IQs that fall in the mild to moderate range of intellectual disability. Children with Down syndrome fully participate in public and private educational programs. Educators and researchers are still discovering the full educational potential of people with Down syndrome.

Myth: *People with Down syndrome have a short life span.*

Truth: Life expectancy for individuals with Down syndrome has increased dramatically in recent years, with the average life expectancy approaching that of peers without Down syndrome.

Myth: *Most children with Down syndrome are born to older parents.*

Truth: Most children with Down syndrome are born to women younger than 35 years old simply because younger women have more children. However, the incidence of births of children with Down syndrome increases with the age of the mother.

Myth: *Most people with Down syndrome are institutionalized.*

Truth: 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. People with Down syndrome are valued members of their families and their communities, contributing to society in a variety of ways.
**Myth:** Parents will not find community support in bringing up their child with Down syndrome.

**Truth:** 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.

**Myth:** Children with Down syndrome must be placed in segregated special education programs.

**Truth:** 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 current trend in education is for full inclusion in the social and educational life of the community. Increasingly, individuals with Down syndrome graduate from high school with regular diplomas, participate in post-secondary academic and college experiences and, in some cases, receive college degrees.

**Myth:** Adults with Down syndrome are unemployable.

**Truth:** 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, childcare, the sports field and in the computer industry. People with Down syndrome bring to their jobs enthusiasm, reliability and dedication.

**Myth:** People with Down syndrome are always happy.

**Truth:** People with Down syndrome have feelings just like everyone else in the population. They experience the full range of emotions. They respond to positive expressions of friendship and they are hurt and upset by inconsiderate behavior.

**Myth:** Adults with Down syndrome are unable to form close interpersonal relationships.

**Truth:** People with Down syndrome date, socialize, form ongoing relationships and marry.

*Sources for this article were publications from the National Down Syndrome Society*