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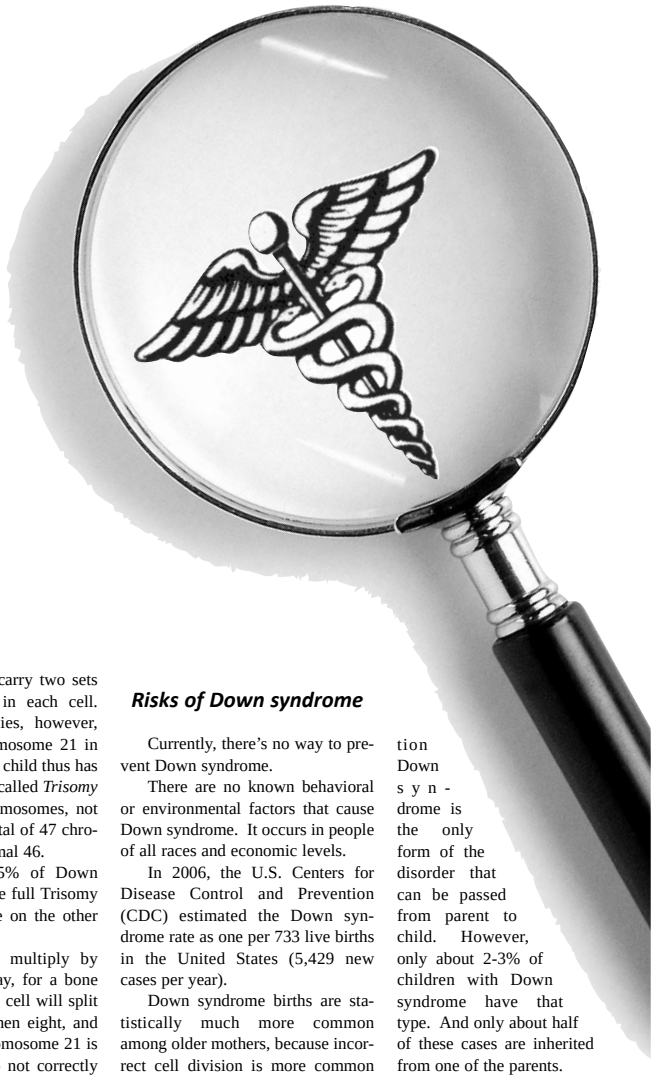
In the Know

All about… Down syndrome



Fear of the unknown, and the misunderstandings and offenses it can engender, is perhaps nowhere more seen than in uninformed reactions to individuals with Down syndrome.

That's because Down syndrome is the world's most common genetic medical disorder. Chances are you've seen its unmistakable signs on the faces of numerous children or young adults.

It also can lead to mistakes—errors in thought, speech or action, many committed with the finest of intentions. But with a little knowledge, sensitivity and compassion, Down syndrome, like any other disability, can easily be demystified.

What is Down syndrome?

Down syndrome is named after Dr. John Langdon Down, the British physician who studied it in the 1860s.

People with Down syndrome have a distinct facial appearance. Though not all people with Down syndrome have the same features, some of the more common ones are flattened nose, protruding tongue, small head and/or chin, upward-slanting eyes, and unusually shaped ears or short neck.

People with Down syndrome may also have hearing deficits, poor muscle tone, short hands with a single crease in the palm, relatively short fingers, excessive flexibility, and a larger-than-normal space between the first and second toes.

Infants born with Down syndrome may be of average size, but typically they grow slower and remain shorter than other children of similar age. Children with Down syndrome also have some degree of mental retardation, from very mild (IQ 50-70) and moderate (IQ 35-50) to severe. Most people with Down syndrome have MR in the mild to moderate range.

Health concerns for individuals with Down syndrome include congenital heart defects, gastroesophageal reflux disease, recurrent ear infections, obstructive sleep apnea, and thyroid dysfunctions.

On the other hand, people with Down syndrome enjoy several as-of-yet unexplained health benefits, including lower risk of hardening of the arteries and diabetic retinopathy, and greatly reduced occurrences of many common cancers except leukemia and testicular cancer.

Due to Dr. Down's perception that children with the syndrome shared facial similarities with the "Mongolian race," Down used the term *Mongoloid* to describe them. Down syndrome individuals were thus described as "having Mongoloid features" or being "Mongoloid" until about a generation ago; that phrase has been largely discarded.

A century ago, Down syndrome children barely lived past age ten. Today, thanks to vast tectonic shifts in societal thinking and major medical breakthroughs, individuals with Down syndrome can live relatively happy, healthy, productive and mainstreamed lives through their 40s and 50s and even beyond.

Causes and types of Down syndrome

In 1959, researcher Jerome Lejeune discovered that Down patients had three, not the normal two, copies of Chromosome 21. Down syndrome was thus found to be genetic in cause.

To preface, the human body contains billions of cells: skin cells, bone cells, muscle cells and so on. Each cell contains 46 chromosomes—two identical sets of 23 each, one from each parent.

The chromosomes tell each cell, and each body part, exactly how to grow. Most physical characteristics, and physical defects, are dictated by precise chromosomal configurations.

Normally, babies carry two sets of 23 chromosomes in each cell. Down syndrome babies, however, carry one extra Chromosome 21 in one of those sets. The child thus has a condition medically called *Trisomy 21*, or three #21 chromosomes, not two. This creates a total of 47 chromosomes, not the normal 46.

Approximately 95% of Down syndrome cases are the full Trisomy 21 incidences. (More on the other 5% below.)

The body's cells multiply by dividing—meaning, say, for a bone to grow, a single bone cell will split into two, then four, then eight, and so on. The extra Chromosome 21 is created when cells do not correctly divide at various points of the fetus' very early development. When a complete third copy of Chromosome 21 is created, either from the father or mother, Trisomy 21 results.

In 1-2% of Down syndrome cases, some cells first contain the extra chromosome but then somehow do not. Alternatively, only some, not all, of the body's cells contain the extra chromosome. These two variants are called Mosaic Down syndrome.

The remaining 2-3% of Down syndrome occurs when extra pieces of Chromosome 21 are created—but become physically *translocated*, or attached to another chromosome, usually Chromosome 14. These are known variously as *Translocation, Robertsonian* or *Familial* Down syndrome.

In extremely rare cases, if the extra copy of Chromosome 21 is missing a few pieces—or consists of only a few pieces—it creates "Partial-copy" Down syndrome, in which the physical and/or mental characteristics of Down syndrome may not even be present.

The medical effects of the extra copy (or pieces of copy) vary greatly, depending on the extent of the extra copy and genetic history.

Risks of Down syndrome

Currently, there's no way to prevent Down syndrome.

There are no known behavioral or environmental factors that cause Down syndrome. It occurs in people of all races and economic levels.

In 2006, the U.S. Centers for Disease Control and Prevention (CDC) estimated the Down syndrome rate as one per 733 live births in the United States (5,429 new cases per year).

Down syndrome births are statistically much more common among older mothers, because incorrect cell division is more common among older mothers. For example, at age 20 to 24, the probability is about one in 1,500, while a 35-year-old has about a one in 350 chance, which increases gradually to about one in 100 by age 40. At age 45, the incidence becomes approximately one in 33 and above age 45, the probability is about one in 19.

Although the probability increases with maternal age, 80% of children with Down syndrome are born to mothers under 35, simply because younger mothers have more babies. Recent data suggest that paternal age, especially beyond 42, also increases Down syndrome risk.

Other factors may also play a role: A mother with one Down syndrome child has about a one in 100 chance of having another, and being a carrier of the genetic translocation for Down syndrome can result in the translocation being passed on to children.

In these translocation cases, the mother or father is a *balanced carrier* of the translocation, which means he or she has some rearranged chromosomes, but no extra pieces of chromosomes. A balanced carrier has no signs or symptoms of Down syndrome, but he or she can pass the translocation on to children.

As a matter of a fact, translocation Down syndrome is the only form of the disorder that can be passed from parent to child. However, only about 2-3% of children with Down syndrome have that type. And only about half of these cases are inherited from one of the parents.

The chance of passing on the translocation depends on the gender of the parent who carries it: If the father is the carrier, the risk is about 3 percent; if the mother is the carrier, the risk is about 12 percent.

Diagnosing Down syndrome

If Down syndrome is not diagnosed before birth, a newborn's physical features often clearly tell doctors whether the child has Down syndrome. But these traits can be subtle in a newborn.

Doctors can often make a diagnosis by doing a physical exam. But to be absolutely certain, doctors will order a blood sample taken for a *chromosomal karyotype*, an analysis of the child's chromosomes.

It may take two to three weeks to get the complete results of this test. If there's an extra Chromosome 21 present in all or some of the cells, the diagnosis is Down syndrome.

Becoming a Down syndrome parent

When parents learn that their child has Down syndrome, they may experience a wide range of emotions, including anger, fear, worry,

sorrow, self-pity and guilt. They may not know what to expect, and may worry about their ability to care for a baby with a disability. This is normal.

However, the best antidote for these legitimate emotions is information and support.

In addition to their child's primary care pediatrician, parents are strongly urged to select a team of trusted professionals, including a pediatric cardiologist, gastroenterologist, endocrinologist and developmental pediatrician. Each of these will help parents understand the various medical problems their infant or toddler may face. They will also help parents evaluate the resources in their area and explain state and federal programs for children with disabilities.

Later, as the child grows, an audiologist, physical therapist, speech therapist, occupational therapist and other specialists will likely be needed to help the child improve to the extent possible.

Most importantly, parents are exhorted to seek out other families who are dealing with the same issues—and to soundly reject any misinformation about Down syndrome.

Some people falsely believe that children with Down syndrome must be placed in segregated special education schools and that adults with Down syndrome usually live in institutions. However, many people with Down syndrome live with their families or independently, go to mainstream schools, read and write, and have jobs. Today, people with Down syndrome can live fulfilling lives.

And today, thanks to ongoing societal progress, most communities have support groups for parents of children with Down syndrome.

Breaking the news

One of the greatest difficulties parents face is finding a way to tell family members and friends about their newborn child's condition. If you do not learn that your baby has Down syndrome until after he or she is born, you will have little time to absorb the information before you need to answer questions from excited family and friends who are eager for news. The best approach may be to simply state the facts, such as, "Our baby was diagnosed with Down syndrome."

There is no right or wrong way to tell people. If you are not ready to talk about your child's condition beyond that, say so.

Hamaspik can put you in touch with other Down syndrome parents and families; contact your Hamaspik MSC or Central Intake. In the meantime, try to focus on caring and bonding with your newborn and getting the help you need. Your doctor or local hospital can also refer you to community resources to help you learn what to expect and how to care for your baby with Down syndrome.

Treatment

Overview

Your child's health team will direct treatment for your child's unique symptoms and physical problems. By working closely with these health professionals and other care providers, you can help your child become as independent as possible and lead a healthy, meaningful life.

Initial treatment

Your newborn with Down syndrome will have routine checkups and various tests during the first month. These tests are used to monitor his or her condition and to help health professionals look for early signs of common diseases associated with Down syndrome and other health problems. These checkups also are a good time to begin discussing issues of concern about your newborn.

Ongoing treatment

Three important parts of ongoing treatment for Down syndrome are 1) making sure that your child has regular medical checkups, 2) helping to manage his or her adjustments to social and physical changes, and 3) promoting independence.

Your child's regular checkups will screen for common and age-group-specific problems seen in individuals with Down syndrome, such as heart, hearing, or vision problems.

From birth to one month, concerns often focus on preventing colds and infections. Also, you might start exploring the types of therapies that might be tried based on how your child develops.

During their first year of life, children with Down syndrome generally meet developmental milestones, but it may take them longer than children without Down syndrome. For example, children with Down syndrome may take twice as long to sit, crawl, walk or talk.

Early intervention programs, which are highly recommended and should be started as soon as possible, may give kids with Down syndrome the best chance of success in life.

From age 1 through 5, concerns are often related to your child's rate of growth and development, which is typically slower than other children of the same age. You will likely want to address behavior, social skills, diet and exercise, and how to prevent common illnesses.

Ages 5 to 13, the middle and late childhood years, see concerns increasingly focused on gaining independence, social skills, and education.

Regarding the 13 to 21 span, adolescent and young adult concerns often relate to the transition into adulthood and planning for the future, such as where your child will live.

Independence and self-sufficiency is influenced by your child's mental and physical abilities. Although it may take extra time for your child to learn and master skills, you may be surprised at how much he or she will be able to do. With proper encouragement and guidance, your child can master all or most major life skills.

Current research

Thanks to continued discoveries, geneticists and other scientists on the front lines of Down syndrome research are able to increasingly focus on the condition's smallest and most detailed inner workings—to discover exactly what causes them, and how to possibly prevent or counteract them in the future.

To elaborate: Science now knows that Down syndrome is caused by an extra Chromosome 21. Chromosome 21 consists of a number of genes. Researchers are therefore working to learn which precise gene causes which symptom of Down syndrome.

For example, at the Center for Down Syndrome Research at Stanford University, teams of scientists are focusing primarily on *cognition*, or the brain's ability to learn, remember and otherwise think—and on which extra Chromosome 21 gene causes the hindered cognition commonly known as "mental retardation." Their hope is to eventually learn how to "turn it off" or otherwise prevent it from triggering reduced cognition.

Specifically, these scientists are focusing on the *hippocampus*, the part of the brain in which cognition occurs. More specifically, they are focusing on the synapses, or the "wires" in the hippocampus that pass information between brain cells, allowing for normal cognition.

At the same time, medical treatments for many of the physical symptoms of Down syndrome, like leukemia, infections and heart problems, are more advanced than ever, allowing individuals with Down syndrome to live longer, healthier lives than ever. Also, speech and other early-intervention therapies are making more of a difference in Down syndrome individuals' lives than ever, allowing them greater opportunities for mainstreaming than ever.

Bottom line? Many professionals may have a hand in helping your child through life. But from birth on, parents play a vital role in their child's success. ■