

# Understanding Down syndrome: A Parent's Guide

## **Introduction**

As a parent, you may have many questions regarding your child's Down syndrome diagnosis. As you look for answers, you may encounter information about Down syndrome, its causes, and possible treatments. All of these different opinions can make it challenging for a family to organize options and begin to choose a treatment plan that best fits the family.

We hope that this packet will help better prepare you to understand the information you receive about treatments for Down syndrome, as well as give you the chance to look over the most recent professional opinions about Down syndrome.

Westside Regional Center anticipates supporting your family as you proceed from your child's initial diagnosis to ongoing support throughout his or her lifetime.

## **What is Down syndrome?**

Down syndrome is one of the most common genetic disorders, affecting about 1 in 800 to 1000 live born children. It occurs among all ethnic groups and economic classes. The disorder is caused by the presence of an extra chromosome – people with Down syndrome are born with *three* copies of chromosome 21, instead of the usual two. This is caused by an error in cell division called “nondisjunction”, which usually occurs at conception and is not related to anything the mother did during pregnancy. Unfortunately, we still do not know why this happens, but we do know that older mothers, such as over age 35 have a higher chance of having a baby with Down syndrome than younger ones.

## **How did my child receive this diagnosis?**

Down syndrome may be diagnosed before or after birth. Several screenings (such as the first trimester and second trimester screens, triple screen or AFP blood tests) may detect high risk during pregnancy. They diagnose using tissue and fluid in the womb around the baby that can be checked for the extra chromosome (called amniocentesis or CVS).

Down syndrome may also be identified at birth or shortly thereafter. Usually, an initial diagnosis is made if the baby has the physical signs of or birth defects associated with Down syndrome.

Some of the most common physical signs of Down syndrome include:

- A slightly flattened facial profile or a flat nose bridge
- Folds at the inner corners of the eyes or an upward slant to the eyes
- Ears that are noticeably small or look lower than usual
- Single crease across the palm of their hand
- Curved “pinky” fingers
- A large gap between the big toe and the second toe

Some of the more noticeable birth defects associated with Down syndrome are:

- Poor muscle tone

- Heart disease
- Gastrointestinal atresia (problems with intestines/digestion)
- Eye disease (eg strabismus (crossing eyes), cataracts, nystagmus, poor vision)
- Hearing problems

However, while the initial diagnosis may be based on some of the physical characteristics listed above, the diagnosis will be confirmed by a chromosome study (usually by examining blood). Studying the chromosomes allows the doctor to confirm whether or not your child has the extra chromosome 21 associated with Down syndrome.

### **Evaluating Treatments**

Before you explore the various treatments for Down syndrome, here are some suggestions to help you evaluate your options for your child's therapy.

**Find a team of trusted professionals.** You'll need to make important decisions about your child's education and treatment. Find a team of doctors, teachers and therapists that you trust. These professionals can help evaluate the resources in your area and help explain the federal regulations regarding children with disabilities. WRC physicians can help you with local resources if desired.

**Seek out other families who are dealing with the same issues.** Most communities have support groups for parents of children with Down syndrome, as well as the option for online forums on the Internet. This can be a great outlet for emotional support, as well as a place to discuss treatment experiences with other parents. The Family Resource and Empowerment Center can provide you with information on family support groups.

**Don't believe the myths about Down syndrome.** Some people believe that children with Down syndrome must be placed in segregated special education schools and that older adults with Down syndrome usually live in institutions. These myths simply aren't true. Many people with Down syndrome live with their families, go to mainstream schools, read and write, and perform various jobs as adults. People with Down syndrome can live fulfilling independent or semi-independent lives.

**Determine whether a treatment is right for your child.** No child with Down syndrome is the same –children with DS often display a wide range of functioning and developmental differences. For example, intelligence for children with Down syndrome can range from average to severe intellectual disability. Similarly, there can be a wide range of functioning due to differences in muscle tone. For these reasons, one of the most important things to remember when thinking about treatments is to plan them specifically for your child. You must ask yourself, *“Is the treatment right for my child? Which issues do I specifically want to address?”* The best place to start is to have a complete assessment of your child's strengths, challenges, and needs by a team of medical professionals, including a physician, physical therapist, occupational therapist, and a speech therapist (by the medical professionals working within WRC or medical professionals that you already have). The exam would allow you to research treatments that specifically focus on the issues that are most important for your child.

**Understand how much time and commitment the treatment will require.** It is important to know that your family will play an important role in supporting and helping to carry out treatments. Therefore, many treatments may be stressful, not only for your child, but for your entire family. Some treatments may not have great benefit for your child in his or her situation. When considering possible treatments, you should consider both the financial and emotional costs. *Does the benefit outweigh the costs for your child and family?* Also, if you decide to pursue a specific treatment, it is important that you know how to decide if the treatment is working. This allows you to monitor the progress of your child, so that you can decide whether or not you should continue or stop the treatment after a period of time.

### **Management and Treatment of Down Syndrome**

Although there is no cure for Down syndrome, there are a variety of treatments that can help make sure your child achieves his or her potential – physically, mentally, and emotionally. Your child’s symptoms can usually improve with early, intensive management, which works on functioning, capabilities, and quality of life.

If your child has Down syndrome, you'll likely become acquainted with a team of doctors that may include a pediatrician, a pediatric cardiologist, a pediatric gastroenterologist, and other pediatric specialists. These doctors can detect and treat complications of Down syndrome, such as heart defects, gastrointestinal problems and hearing problems.

Your child's care team may also include a physical therapist, speech pathologist, occupational therapist and others. These specialists can help your child develop skills as fully as possible. For example, babies with Down syndrome do not usually have good muscle tone, so a physical therapist can help your baby learn to roll over, sit up and walk. Meanwhile, a speech pathologist or occupational therapist may help with feeding, hand coordination and language skills.

In general, children with Down syndrome usually meet developmental milestones, but it may take them a little longer than it does for children without Down syndrome. For example, children with Down syndrome may take twice as long to sit, crawl, walk or say a first word. However, early intervention programs may give children with Down syndrome a more support to meet milestones. You may ask your doctor and your service coordinator about specific early intervention programs designed to help stimulate children with Down syndrome at an early age with appropriate sensory, motor and cognitive activities.

### **What to expect medically**

Individuals with Down syndrome have the same basic healthcare needs as other people, including vaccine schedules, as well as screenings for high blood pressure, heart disease, cancer and dental disease. Your child’s primary doctor should continue to provide your child with regularly-scheduled medical care and preventive health screenings. However, people with Down syndrome are at higher risk for certain medical complications. Your

child may have some of these or may develop them over time, but not every person with Down syndrome has these problems. They can include:

- ***Heart Defects*** – Approximately 30 to 60% of children with DS are born with some form of heart defect and surgery, frequently during the first 6 months of life, may be necessary to prevent serious complications. Therefore, it is extremely important that all babies who might have Down syndrome be evaluated for heart defects as early as possible.
- ***Feeding challenges*** – Breastfeeding is beneficial for infants with DS as it enhances oral motor development and increases bonding between mother and child. However, sucking problems related to cardiac defects or low muscle tone (hypotonia) may result in feeding difficulties. Please consult with a feeding specialist/lactation nurse if your child has any problems with feeding.
- ***Vision and hearing problems*** – Many individuals with DS also have problems with hearing and vision. Hearing loss can be related to fluid buildup in the inner ear or to structural problems of the ear itself. Vision problems commonly include amblyopia (lazy eye), near- or farsightedness, and an increased risk of cataracts. *Yearly hearing evaluations by an audiologist (hearing specialist) or an otolaryngologist (ear, nose and throat doctor) and an ophthalmologist (eye doctor) are necessary to detect and correct any problems before they affect your child's language and learning skills.*
- ***Thyroid disease*** – Individuals with DS have an increased risk of developing thyroid disease, usually hypothyroidism. Thyroid disease occurs when the thyroid gland does not supply the proper amount of hormones needed to help regulate metabolism and growth. Children with hypothyroidism might seem sluggish. They might gain weight, even though they're not eating more or getting less exercise than usual. Teens with hypothyroidism also might have slow growth in height, slow sexual development, irregular menstrual periods in girls, muscle weakness, dry skin, hair loss, poor memory, depression and difficulty concentrating. *All individuals with Down syndrome should be tested for hypothyroidism at birth and at least every year thereafter.* In particular, as the thyroid hormone affects normal development of the brain, testing of infants is particularly crucial.
- ***Dental problems*** – People with Down syndrome are at greater risk for gingivitis and periodontal disease. It is extremely important to promote a diet that does not increase the risk of tooth decay, in addition to establishing a daily oral hygiene routine that focuses on plaque removal by regular brushing and flossing. *It is also very important for your child to have their dental visits twice a year so that the dentist can monitor your child's gums and teeth before any serious infections occur that might cause tooth loss.*

- **Low muscle tone** – Children with Down syndrome have an increased risk of low muscle tone (hypotonia). Some common signs of this include motor skills delay (such as a delay in the ability to roll, sit up, crawl, or walk), hypermobile or very flexible joints, drooling and speech difficulties, poor reflexes, decreased strength, decreased activity tolerance, or rounded shoulder posture. Low muscle tone may also affect the muscles of the digestive system, in which case constipation may be a problem. While your child may continue to have some muscle weakness, physical therapy can help improve motor control and overall body strength. Also, occupational therapy can help develop fine motor skill development and hand control, and speech-language therapy can help with breathing, speech, and swallowing difficulties.
- **Slow/delayed growth & development** – It is common for a child with Down syndrome to experience slow or delayed growth and development. For example, it may take your child longer to learn how to turn over, sit, stand, and respond. However, while the development of speech and language abilities may take longer than expected and may not occur as fully as parents would like, your child will likely be able to develop the communication skills they need through early interventions, physical therapy and speech therapy.
- **Leukemia.** Young children with Down syndrome are more likely to develop leukemia than are children who do not have Down syndrome. A complete blood count should be obtained to make sure that your child does not have leukemia.
- **Other diseases.** Because of abnormalities in their immune systems, those with Down syndrome may be more susceptible to infectious diseases. For example, the risk of contracting pneumonia and ear infections is much higher than that of others without Down syndrome. Also, people with Down syndrome are more likely to experience sleep problems such as obstructive sleep apnea, which have symptoms like snoring, restless sleep, and unusual sleep positioning. Additionally, very dry skin and other skin problems are common in people with Down syndrome. Talk with your doctor about optimizing your child's health.
- **Dementia.** Later in life, people with Down syndrome have an increased risk of dementia or Alzheimer's disease. Signs of dementia often appear before age 40 in people with Down syndrome. Early symptoms include loss of memory and logical thinking; personality change; and decline in daily living skills. If you notice any of the above signs, be sure to bring it to the attention of your doctor, so that he/she can monitor your child's condition.

As people with Down syndrome get older, they have a higher risk for some age-related conditions. You and your doctors should watch especially for signs of diabetes and mental illness. People with Down syndrome may have difficulty communicating that they are having problems, such as pain. As family members, you can look out for signs of pain and point these out to your doctor.

Many of the conditions experienced by people with Down syndrome can be avoided or minimized with good medical care, exercise and activity, equipment use, and therapies. Working with your doctor, service coordinator and other professionals can provide excellent medical management, education, and work/job training that can markedly improve the level of functioning of children with Down syndrome so that in adulthood, they may live independent or semi-independent fulfilled lives.

### **Resources at WRC**

- Health and Medical Unit
- Equipment Clinic
- Family Resource Center
- Service Coordination
- Support Groups – Birth to Five Parent, Teen Social Group, Networking and Social Group
- Trainings & Events – self advocacy, consumer discussion, and behavior management

### **Resources in the Community**

American Academy of Pediatrics: [www.aap.org](http://www.aap.org)

American Academy of Family Physicians: [www.familydoctor.org](http://www.familydoctor.org)

National Down syndrome Society (NDSS): 1-800-221-4602 or [www.ndss.org](http://www.ndss.org)

National Down syndrome Congress (NDSC): 1-800-232-6372 or [www.ndscenter.org](http://www.ndscenter.org)

National Parent to Parent Support and Information System: 1-800-651-1151

Down Syndrome Association of Los Angeles (DSALA): [www.dsala.org](http://www.dsala.org)

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