Developmental Expectations and Medical Issues for Children with Down Syndrome

NATIONAL DOWN SYNDROME CONGRESS

Introduction to Genetics

All human beings have 23 unique pairs of chromosomes in each of their cells. Chromosomes carry our genetic information (our genes), which give each of us our initial blueprints (like whether we will be tall or have brown hairor have blue eyes, for example. For most people, we inherit one copy of each chromosome from our parents, resulting in 46 total chromosomes. People with Down syndrome are born with an extra 21st chromosome. Nobody knows for sure why it happens, but we know that the extra chromosome is usually present in every one of their trillions of cells. This extra chromosome can have can potentially lead to different medical or developmental challenges, but can also lead to a lot of positive outcomes as well.

Developmental Expectations and Milestones

Children with Down syndrome typically have some delays in the different areas of development. Gross motor delays are delays in areas like sitting, crawling, and walking. Fine motor delays are delays in purposeful coordination of the movements of the hands and fingers. Speech delays are delays in the onset and quality of speech development. All children develop at a different pace and some may not have as many delays as others in these areas of development. On average, most children with Down syndrome reach their developmental milestones about 1.5 to 2 times later than other children. It is important to remember that almost all children with Down syndrome reach the developmental milestones. We just have to be patient while waiting.

Developmental Milestones

Milestone	Typical Child average (months)	Child with Down Syndrome average (months)
Smiling	1	2
Rolling Over	5	8
Sitting Alone	7	10
Crawling	8	12
Talking (words)	10	16
Standing	11	20
Walking	13	24
Talking (phrases)	21	28

Developmental Skills

Skill	Typical Child average (months)	Child with Down Syndrome average (months)
Finger Feeding	8	12
Using spoon/fork	13	20
Bowel Control	29	42
Bladder control	32	48
Undressing self	32	40
Dressing self	47	58

To help them reach the developmental milestones and skills effectively, it is important to involve early intervention services. These services are provided by each state and involve a variety of different types of therapists including developmental therapists (general development), physical therapists (gross motor), occupational therapists (fine motor), and speech therapists (language and communication skills). Many states also have dieticians and feeding specialists in early intervention. These programs are government funded and are offered through the individual counties in each state. The services are provided from birth to age 3 years. After that, the local school districts take over the therapies.

Medical Issues

There are certain medical conditions that occur more commonly in people with Down syndrome. In the mid-1990s, the first set of Healthcare Guidelines were published, and they were most recently updated in 2011 and re-confirmed in 2016. The goal of these guidelines is to help identify medical issues and concerns before they become more problematic. What follows below is an overview of these medical issues and any associated healthcare guidelines. Please remember that this is only an overview of potential issues that *could* develop and does not mean that every child will have all of these problems. The vast majority of children with Down syndrome are indeed quite healthy and thriving.

Cardiac - Newborn infants with Down syndrome are born with a heart condition about half of the time. Most commonly, the issue is with the structure of the heart and results in mixing of the blood containing oxygen with the blood that is low in oxygen. Some of the heart conditions will require surgery, but many can improve on their own with time and close monitoring. It is important that all children born with Down syndrome have an echocardiogram (an ultrasound of the heart) within the first 1 to 2 months after being born. If there are any abnormalities on the echocardiogram, they should be seen by a pediatric cardiologist. After the newborn period, some adolescents and adults with Down syndrome can develop leaky heart valves that may result in a heart murmur that was not previously present. If this happens, a cardiologist should be consulted.

Gastrointestinal (GI) - About 5% of infants with Down syndrome will be born with an obstruction of their intestinal tract. This can result in feeding problems, vomiting, or significant constipation soon after being born. The newborn nursery medical team should consider one of these possible obstructions if there are any of these symptoms in an infant with Down syndrome. If needed, pediatric surgeons can become involved to address the obstruction. After the newborn period, gastrointestinal issues that are seen include reflux, constipation, celiac disease, or chewing/swallowing difficulties. If any of these issues become problematic, specialists in gastroenterology are often involved. Routine screening for celiac disease in asymptomatic individuals with Down syndrome is no longer recommended.

Respiratory/Ear, Nose, Throat - Many people with Down syndrome have what is called “mid-face hypoplasia.” This simply means that the middle part of their face may be narrowed. This can result in difficulty draining the fluids in that part of the face. Therefore, they can develop recurrent ear infections, recurrent sinus infections, recurrent croup, and sometimes hearing loss. Regular hearing screenings are recommended for people with Down syndrome, beginning at birth. After that, they should be re-screened by an Audiologist every 6 months for the first 3 to 4 years of life to make sure their hearing is adequate for speech development through those years.

After about 4 years of age, they can transition to annual hearing screenings. If they develop any of the recurrent ear, nose, and throat issues as discussed above, they should be seen by an ENT (otolaryngology) specialist.

Another common issue related to the airway narrowing seen in people with Down syndrome is obstructive sleep apnea (OSA). Many people with Down syndrome have OSA which can present with snoring, mouth breathing, abnormal sleep positioning, or restless sleep. It can also be completely without symptoms! Therefore, all children with Down syndrome should have a sleep study performed at a Pediatric Sleep Center by 4 years of age. If sleep apnea goes untreated, it can result in higher blood pressures in their lungs, poor school performance, behavioral problems, or even poor growth. If there is sleep apnea, an ENT (otolaryngology) specialist should be involved.

Ophthalmologic (Eye) - Children with Down syndrome often can have difficulty with their vision that can develop over time. The guidelines recommend having an Ophthalmologist exam done at 6 months and 12 months of age, then annually until 5 years of age, every 2 years from 5 to 12 years of age, and every 3 years after that.

Endocrinologic - Children with Down syndrome can be born with hypothyroidism (underactive thyroid). This should be detected on the newborn screen if they were born in a US hospital. They can also develop hypothyroidism later in life. The guidelines call for labs to check for thyroid function at 6 months and 12 months of age, and then annually for the rest of their life. If hypothyroidism is caught early, it can be easily managed with a once a day thyroid medication. Many doctors will refer to a pediatric endocrinologist for management of hypothyroidism. People with Down syndrome can also develop hyperthyroidism (overactive thyroid) and type 1 diabetes at a higher rate than others.

Hematologic - Leukemia is a possible medical issue that parents tend to be understandably very concerned about. It occurs in 1% of children with Down syndrome. Remember that this means that 99% of children with Down syndrome do not get leukemia. The symptoms would include unexplained fevers, weight loss, abnormal paleness, bleeding, or bruising. If those symptoms were seen, your doctor would send a blood count to evaluate and refer to a Pediatric Hematology/Oncology specialist. Some babies can be born with a type of leukemia called transient myeloproliferative disease (TMD). Therefore, all babies with Down syndrome should have a blood count sent at birth.

Iron deficiency anemia is more common in people with Down syndrome. They should get annual screenings for anemia beginning at 12 months of age.

Musculoskeletal - Babies with Down syndrome have very low muscle tone typically at birth. This low tone continues to be present in their ligaments and joints as they get older. One joint that can be affected commonly is the ankle joint. Due to the lower tone, children with Down syndrome can sometimes be seen walking with flat feet with their ankles turned outward. Orthotics can be used to help stabilize the ankles if that occurs.

The lower tone can also affect a very important joint in the upper neck, just below the head, and result in atlanto-axial instability. The instability can rarely result in spinal cord compression. The symptoms of this instability are neck pain, neck stiffness, sudden arm/leg weakness, loss of bowel/bladder control, change in the way they walk, and abnormal strength/reflexes on the doctor's exam. If any of these symptoms occur, the

children should have immediate evaluation with a neck x-ray and possible referral to a neurosurgeon or orthopedic surgeon, depending on the referral pattern for this condition in the region. To protect against the development of any symptomatic atlantoaxial instability, it is recommended that children with Down syndrome be protected from activities that could result in uncontrolled falls from heights (unattended horseback riding, trampoline use, etc.). Routine screening with a cervical spine x-ray in asymptomatic individuals with Down syndrome is no longer recommended.

Neurologic - Seizures are more common in people with Down syndrome, occurring in about 5-10% of the population, primarily in infancy or in later adulthood. If they were to be suspected, a neurology evaluation would be indicated.

Dental – Children with Down syndrome tend to have delayed eruption of their teeth. Sometimes, the first tooth may not be present until 18 to 24 months of age. The teeth also erupt in an atypical pattern with the first teeth present sometimes being the molars or canines. There can be crowding of the teeth as well. A dentist should start seeing the children as early as the first tooth eruption but at least by 2 to 3 years of age.

General Pediatric Care – Immunizations should be given to children with Down syndrome at the same schedule as for all children. Additional vaccines that may be needed include the respiratory syncytial virus (RSV) vaccine, also called Synagis, if the baby has a cardiac issue that is not repaired or was born significantly premature. Also, children with Down syndrome with any chronic cardiac or respiratory issue should receive the Pneumovax (PPS-23) vaccine once after 2 years of age.

Medical Benefits in Down syndrome

There are several apparent medical benefits of having an extra 21st chromosome that are not frequently discussed. People with Down syndrome very rarely develop solid organ cancers. This means that they do not seem to get lung cancers, liver cancers, brain cancers, etc. It is not known why this is so, but there must be something protective on the 21st chromosome from these types of cancers. They also do not seem to get cholesterol plaques in the arteries around their heart or high blood pressure. Their incidence of dental cavities seems to be less than other children as well. If the children do develop leukemia, their response to chemotherapy is much better in certain types of leukemia. Their response to treatment for specific childhood seizure conditions also seems to be better than other children.

People with Down syndrome also seem to have benefits in their visual memory skills. Many of them are able to recall specific life situations much more clearly than others. They may remember specifics from their childhood birthday parties or the names of acquaintances much better than others. These visual memory skills can even help them excel with reading skills if their visual skills are used effectively.

Conclusion

The vast majority of children with Down syndrome do very well in life. With the appropriate supports and medical screenings, the life expectancy for people with Down syndrome has climbed from 15 years to 65 years over the past several decades.

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