Health Care Information for Families of Children with Down Syndrome

American Academy of Pediatrics

Introduction

Down syndrome is a common condition caused by having “extra” copies of genes on the 21st chromosome. Those extra genes change development during pregnancy, and they continue to have effects after birth and throughout a person’s life. Each person with Down syndrome is unique, having some of the many possible health, learning, and related differences that can occur with this condition.

Some of the differences in people with Down syndrome are common and visible, like the facial appearance. Other changes are less common or less visible but can still cause problems or may need special treatments. The “special treatments” may include medicines, surgeries, or changes in what you should expect. There are no medicines or therapies that are needed by all people with Down syndrome. There are also no medicines or therapies that can “cure” Down syndrome.

Your child’s doctor should be your starting point. Your child needs regular doctor visits and a few special tests. Medical specialists may also need to be involved.

The medical issues for a child with Down syndrome change with age. For this reason, this document is divided into several age groups. Each age group includes a list of issues that may be important to your child at that age. Your doctor can check the full AAP guideline for more details (the web address is given below).

The information within each age group is sorted by the parts of the body that are affected (heart, ears, etc). Many tests only need to be done once. Some areas might need to be looked at again, or even many times, as the child grows to an adult.

This document focuses on medical topics that affect physical health. Other issues can affect social and school success, which may not need doctors or other medical resources but are still important issues for children with Down syndrome. Many people with Down syndrome understand more than they can say. They may need help to communicate in other ways. Most have good social skills, especially if they have friends with typical behavior as models. Respect for and attention to their abilities are often important missing pieces and may be enough to make a big difference in performance and behavior.

The information in these guidelines has grown with the help of families, Down syndrome clinics, and doctors around the world. Most of the information is easy to follow. However, some tests or specialists might be needed that are not available in your area. Your doctor can help to sort out the best next-steps when something can’t be done quickly or nearby.

When you visit the doctor, you might want to bring a notebook to write down information from each visit. A notebook will help you keep all of your child’s medical information in one place. This will be valuable when you meet with new doctors or with others involved in your child’s care.

Local parent support groups can be a very good place to learn about doctors, therapists, and other providers in your community. They may also be able to help with questions about daycare, preschools and schools, other local developmental programs, problems with behavior, help with child care, etc. Your doctor’s office should have names and contact information for groups in your area.
“Health Care Information for Families of Children with Down Syndrome” was created in July 2013 by the American Academy of Pediatrics (AAP) to give parents and families information about the special health care needs for children with Down syndrome. This document should be used together with the care given by a child’s doctor.

This information is based on the “Health Supervision for Children with Down Syndrome” clinical report from the American Academy of Pediatrics, available here: http://pediatrics.aappublications.org/content/128/2/393.full

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Health Care Information for Families of Children with Down Syndrome

Child’s Age: The Prenatal Period (the time before birth)

☐ **Consider testing as desired**

Prenatal testing for genetic conditions is recommended for families who wish information to help them make decisions about a pregnancy. This testing should be done only after information about the tests has been discussed between the doctor and the family, and the family understands the risks and benefits of the testing.

☐ **Counseling**

If Down syndrome (trisomy 21) or any other chromosome change that causes Down syndrome is found by prenatal testing, the family should receive counseling to explain the issues and provide support for the family.

☐ **Prenatal heart testing**

Because there is a high risk of heart problems at birth in Down syndrome, echocardiography (an ultrasound picture of the heart) done during the pregnancy can provide information that may be useful for the remainder of the pregnancy and for the delivery. This information may help with decisions such as where to deliver the baby and the medical services needed late in pregnancy or at delivery.
Health Care Information for Families of Children with Down Syndrome

Child’s Age: Birth to 1 Month

☐ **Complete physical examination**
   If the diagnosis of Down syndrome was made before birth or suspected after birth, a complete physical examination should be done to confirm the known physical features and to check for any possible associated conditions.

☐ **Genetic testing**
   If prenatal testing gave a diagnosis of Down syndrome and if the exam after birth agrees, then no further testing is probably needed in the newborn period. If the physical examination after birth raises the possibility of Down syndrome, testing by rapid (FISH) confirmation and a complete chromosome analysis are needed. The rapid analysis results are typically available within 48 hours, whereas the complete analysis might take 3-5 days for the results. A complete chromosome analysis is needed to provide full information, but to ensure prompt results, both should be obtained unless the complete analysis can be done as quickly as the rapid analysis.

☐ **Counseling**
   The prenatal or newborn diagnosis of Down syndrome can cause many concerns for parents. Talking with a medical genetics team (medical geneticist and genetic counselor) or others recommended by your child’s doctor may be helpful.

☐ **Feeding**
   Infants with Down syndrome sometimes have low muscle control, which can cause feeding problems. For this reason, infants should be closely watched for slow feeding or choking and for good weight gain. Breastfeeding is strongly encouraged, but extra attention may need to be given to positioning and to keeping the baby awake or alert.

☐ **Heart**
   An echocardiogram (an ultrasound picture of the heart) is needed to check for any evidence of heart disease. This should be done even if a prenatal echocardiogram was done. If issues exist, it is very important to act early. Breathing that is too fast or cyanosis (a bluish color of the skin) are signs for possible concern.

☐ **Hearing and vision**
   Infants with Down syndrome are at risk for sensory issues, such as eye problems leading to vision loss or ear problems leading to hearing loss. It is important to have both vision and hearing checked by specialists (ophthalmology and ENT).

☐ **Thyroid**
   Thyroid hormone levels can be too low in newborns and need to be checked (a TSH test). Thyroid hormone imbalance can cause a variety of problems that might not be easy to detect without a blood test.

☐ **Blood test**
   After birth, white and red blood counts can be unusually high in infants with Down syndrome. These blood counts need to be checked.

☐ **Stomach or bowel problems (reflux, constipation, blockages)**
   Intestinal issues can occur. Spitting up, stomach swelling, or an abnormal stool pattern can be signs that there is an issue.

☐ **Infection**
   Because of an increased risk of infections (especially respiratory infections), infants should be protected from any unnecessary exposures to sick siblings, relatives, or others. It is also recommended to get checked quickly when any infection is suspected.

☐ **Developmental services**
   It is not too early in the first month of life to start to look for the developmental services (sometimes called “Early Intervention”) that will be very important in early childhood.

☐ **Resources**
   Families of children with Down syndrome will need multiple resources, and now is a good time to start lining them up. Such resources might include specialized medical care, early intervention, physical therapy, and family counseling services.
Health Care Information for Families of Children with Down Syndrome

Child’s Age: 1 Month to 1 Year

☐ Regular well-care visits (check-ups)
While infants with Down syndrome might need multiple special visits to their doctor and specialty physicians, it is very important that they get regular well-care visits (check-ups). These visits will include checking your child’s health, giving immunizations (shots), and building the relationships between the doctor and the family. Developing these relationships will help support the medical and other needs of the child and the family.

☐ Monitor growth
It is important to check growth at every visit. Measurements include height, weight, weight for height, and head circumference. Discuss your child’s diet, activity level, bowel and urine patterns, and growth. Your child’s doctor can help with questions about any need for vitamins or supplements.

☐ Immunizations (shots)
Your child’s doctor should follow the same shot schedule as for any other child. This includes yearly influenza (flu) shots. It may include other shots, too, depending on your child’s health history.

☐ Heart
If there were any signs of heart disease in the first month of life, heart monitoring is probably already in place. Heart problems could still worsen or new ones could arise. If concerns exist, it is very important to act early. Breathing that is too fast or cyanosis (a bluish color of the skin) are signs for possible concern.

☐ Hearing and vision
Infants with Down syndrome are at risk for eye problems leading to vision loss or ear problems leading to hearing loss. It is important to have both vision and hearing checked by specialists (ophthalmologist and otolaryngologist/ear, nose, and throat doctor or ENT). The eyes should be tested at birth and again at 1 year or sooner if there are concerns. Hearing should be tested at birth and again every 6 months in early childhood to be sure that the baby’s hearing is the best possible.

☐ Thyroid
Thyroid hormone levels can be too low in infants and need to be checked (a TSH test). Low thyroid levels can cause a variety of problems that might not be easy to detect without a blood test. A TSH should be obtained at birth and again at age 6 months and 1 year.

☐ Stomach or bowel problems (reflux, constipation, blockages)
Intestinal issues can occur. Spitting up, stomach swelling, or an abnormal stool pattern can be signs that there is an issue.

☐ Neck instability
Bones in the neck or spine can be unstable in some people with Down syndrome. There are almost always visible signs when there are problems. Daily physical activity is important to your child and should not be limited by unneeded worries. X-rays are not needed unless there is pain or changes in the use of hands, walking, or bowel or bladder function. If x-rays are done and the results are abnormal, your child may be referred to a spine or neck specialist. It is recommended that the neck be positioned properly for any medical procedures.

☐ Developmental services
Developmental services (for example, early intervention programs) can be of great benefit to the family with a child with Down syndrome. Developmental services can also help arrange for other related services. These services should provide information to your child’s doctor to maintain a close working relationship with the doctor and the family.

☐ Social support services
Many families need additional help with the issues that can arise with the care of children with Down syndrome. All families should discuss with their doctor the social services that may be available and their benefits.

☐ Recurrence risk counseling
Families should get counseling about the possible risk of having another child with Down syndrome, if they choose to have more children. While the risk is usually low, other factors in the family history might be present, so counseling should be done after a complete review of the family history.
Health Care Information for Families of Children with Down Syndrome

Child's Age: 1 Year to 5 Years

☐ **Regular well-care visits (check-ups)**
At the one-year check-up, you should look at the checklists for newborns and infants to be sure everything has been done as recommended. Follow-up on known problems with specialists and be sure that reports are sent to your child’s primary doctor.

☐ **Monitor growth**
It is important to check growth at every visit. Measurements include height, weight, body mass index (BMI), and head circumference. Discuss your child’s diet, activity level, and growth. Your child’s doctor can help with questions about any need for vitamins or supplements.

☐ **Immunizations (shots)**
Your child’s doctor should follow the same shot schedule as for any other child. This includes yearly influenza (flu) shots. It may include other shots, too, depending on your child’s health history.

☐ **Heart**
The need to see a cardiologist during this age is based on the child’s health history and examination. Children with cardiac lesions may need to be monitored even after repair for remaining lesions and development of pulmonary hypertension (high pressure in blood vessels of the lungs).

☐ **Hearing**
Hearing should be checked every 6 months, with audiogram and tympanometry tests until normal hearing is documented by testing of both ears separately (usually by 4-6 years of age). Children with hearing loss should be referred to an otolaryngologist (ear, nose, and throat doctor or ENT). Higher risks of hearing problems can go with middle ear fluid and ear infections. Treatment of middle ear fluid often includes the use of ear tubes.

☐ **Vision**
Vision should be checked at each visit to the doctor and with yearly checkups by a pediatric ophthalmologist (special eye doctor) or a general ophthalmologist who is good with children with disabilities. Crossing eyes or blocked tear ducts might be reasons for quicker action. Early use of eye patches, glasses, or both may help to fix eye crossing while lowering the need for surgery and the risk of vision loss.

☐ **Thyroid**
The thyroid gland is usually normal in babies with Down syndrome. It can stop working normally for half of people with Down syndrome by adulthood. The symptoms of low thyroid can be hard to notice in people with Down syndrome, so a blood test (TSH) is needed every year, or sooner if symptoms change. When there is a problem, treatment is safe and can often be started by your primary doctor.

☐ **Blood tests**
Tests for low iron or anemia (hemoglobin and other tests if needed) should be done every year.

☐ **Stomach or bowel problems (diarrhea, constipation)**
Discuss toilet patterns at each visit, especially any ongoing problems with loose stools or constipation. These are common in children with Down syndrome. Some children with Down syndrome have celiac disease, which is a problem with tolerating some grains, including wheat. Testing can help to identify that condition, and may lead to changes in diet. Celiac disease can affect growth, stooling patterns, and behavior. Let your child’s doctor know if your child is having:
- Very loose stools
- Hard to treat constipation (hard or painful stools)
- Slow growth/weight loss
- Belly pain or stomach swelling
- New or challenging behavior problems
☐ **Neck instability**
Bones in the neck or spine can be unstable in some people with Down syndrome. There are almost always visible signs when there are problems. Daily physical activity is important to your child and should not be limited by unneeded worries. X-rays are not needed unless there is pain or changes in the use of hands, walking, or bowel or bladder function. If x-rays are done, and the results are abnormal, your child may be referred to a spine or neck specialist. Special neck positioning may be needed for some medical procedures. Let your child’s doctor know if your child is having:

- Stiff or sore neck
- Change in stool or urination pattern
- Change in walking
- Change in use of arms or legs
- Numbness (loss of normal feeling) or tingling in arms or legs
- Head tilt

☐ **Sleep issues**
Obstructive sleep apnea is a common problem for people with Down syndrome, especially those with low muscle tone. Some symptoms are obvious (snoring, restless waking at night, daytime sleepiness), but it can be hard to tell just by watching. AAP guidelines recommend that every child with Down syndrome have a sleep study by the age of 4 years. (That testing may be hard to find in some parts of the country.) Treatment can include special breathing equipment or surgery.

☐ **Skin**
Discuss with your child’s doctor if your child has very dry skin or other skin problems.

☐ **Brain and nervous system**
Discuss with your child’s doctor concerns about neurologic problems, such as seizures.

☐ **Dental**
Delayed and missing teeth are common. Teeth often come in unusual order.

☐ **New treatments**
Talk to your doctor about any new treatments or medications you may consider.

☐ **Recurrence risk counseling**
Talk to your doctor about future pregnancy planning and chances of recurrence of Down syndrome and where prenatal diagnosis is available.

☐ **Developmental services (early intervention)**
Review your child’s development with your doctor. Referral to local early intervention services and other options for therapy may be needed. Speech progress can be very delayed in children with Down syndrome, but after some delays, most will learn to talk well. Until speech is easier for your child, he or she might need help finding other ways to communicate, such as using sign language, pictures, reading, or using electronic communication tools. Behavior problems are often linked to problems with communication, but may reflect other issues, including ADHD or autism. Language delays or hidden abuse are more common than autism but may be misdiagnosed. Talk with your doctor about how to explain social safety and “good and bad touch” as your child grows older.
Health Care Information for Families of Children with Down Syndrome

Child’s Age: 5 to 13 Years

☐ Regular well-care visits (check-ups)
   It is important to have yearly well-care check-ups. These visits will assist in checking your child’s health, giving shots, and answering your questions about your child’s health.

☐ Monitor growth
   It is important to check growth at every visit. Measurements include height, weight, and body mass index (BMI). Discuss your child’s diet, activity level, and growth. Your child’s doctor can help with questions about any need for vitamins or supplements.

☐ Immunizations (shots)
   Your child’s doctor should follow the same shot schedule as for any other child. This includes yearly influenza (flu) shots. It may include other shots, too, depending on your child’s health history.

☐ Heart
   The need to see a cardiologist (heart doctor) during this age is based upon a child’s health history and examination. About half of children with Down syndrome are born with differences in how their heart is formed. Children with normal newborn testing for heart problems may not need more evaluation at this age.

☐ Hearing
   Hearing testing is needed every 6 months, until each ear can be tested alone. When a child can respond to testing in each ear alone, testing may be done every year. Children with hearing loss should be referred to an otolaryngologist (ear, nose, and throat doctor or ENT).

☐ Vision
   Vision should be checked at each well-care visit. It is also important to have an exam at least every 2 years by a pediatric ophthalmologist (special eye doctor) or a general ophthalmologist who is good with children with disabilities. Visits may be needed more often if your child has known eye or vision issues.

☐ Thyroid
   The thyroid gland is usually normal in babies with Down syndrome. It can stop working normally for half of people with Down syndrome by adulthood. The symptoms of low thyroid can be hard to notice in people with Down syndrome, so a blood test (TSH) is needed every year, or sooner if symptoms change. When there is a problem, treatment is safe and can often be started by your primary doctor.

☐ Blood tests
   Tests for low iron or anemia (hemoglobin and other tests if needed) should be done every year.

☐ Stomach or bowel problems (diarrhea, constipation)
   Discuss toilet patterns at each visit, especially any ongoing problems with loose stools or constipation. These are common in children with Down syndrome. Some children with Down syndrome have celiac disease, which is a problem with tolerating some grains, including wheat. Testing can help to identify that condition, and may lead to changes in diet. Celiac disease can affect growth, stooling patterns, and behavior. Let your child’s doctor know if your child is having:
   • Very loose stools
   • Hard to treat constipation (hard or painful stools)
   • Slow growth/weight loss
   • Belly pain or stomach swelling
   • New or challenging behavior problems
☐ **Neck instability**
Bones in the neck or spine can be unstable in some people with Down syndrome. There are almost always visible signs when there are problems. Daily physical activity is important to your child and should not be limited by unneeded worries. X-rays are not needed unless there is pain or changes in function. If x-rays are done, and the results are abnormal, your child may be referred to a spine or neck specialist. Special neck positioning may be needed for some medical procedures. Let your child’s doctor know if your child is having:

- Stiff or sore neck
- Change in stool or urination pattern
- Change in use of arms or legs
- Head tilt
- Change in walking
- Numbness (loss of feeling) or tingling in arms or legs

☐ **Sleep issues**
Obstructive sleep apnea is a common problem for people with Down syndrome. This is a sleep problem that can affect a child’s behavior and ability to pay attention. It can also affect the heart. Some symptoms are obvious (snoring, restless sleep, waking at night, daytime sleepiness), but it can be hard to tell just by watching. AAP guidelines recommend that every child with Down syndrome have a sleep study by the age of 4 years. (That testing may be hard to find in some parts of the country.) Let your child’s doctor know if your child is having:

- Loud breathing
- Snoring
- Waking up often at night
- Daytime sleepiness
- Restless sleep (moving around a lot)
- Uncommon sleep positions (like sitting up to sleep or sleeping with neck arched back)
- Pauses in breathing during sleep
- Behavior problems

☐ **Skin**
Discuss with your child’s doctor if your child has very dry skin or other skin problems.

☐ **Brain and nervous system**
Discuss with your child’s doctor concerns about neurologic problems, such as seizures.

☐ **Behavior and mental health**
Some children with Down syndrome can have behavior or mental health issues that affect how they play and work at home, at school, or in the community. Let your child’s doctor know if your child is having problems that make it hard for them to function in the home, community, or school, including:

- Anxiety
- High activity level
- Wandering off
- Not listening
- Other behavior concerns
- Attention problems
- Sad mood or lack of interest in activities
- Loss of skills (not being able to do the things they used to do)
- Obsessive compulsive behaviors (behaviors that are repeated frequently)

☐ **Development**
As for all children, your child’s well child visit is a chance to discuss his/her development, including:

- **School progress and development:** discuss your child’s development, school placement, and service needs
- **Transition:** Children can need extra support at times of transition: discuss transition from elementary to middle school
- **Social/independence skills:** discuss developing social skills, self-help skills, and a sense of responsibility. Discuss working toward independence (doing things by him- or herself) with bathing, grooming, and self-care

☐ **Sexuality and puberty**
Children need help to learn appropriate touch in social situations and correct names for body parts. Puberty can bring many changes. As your child approaches and enters puberty, discussions can include:

- Changes in puberty
- Managing sexual behaviors (such as masturbation)
- Gynecologic (women’s health) care for girls with Down syndrome, including periods and mood changes related to (menstrual) periods.
- Fertility, birth control, prevention of sexually transmitted infections
- Risk for a person with Down syndrome having a child with Down syndrome
Health Care Information for Families of Children with Down Syndrome

Child’s Age: 13 to 21 Years or Older

☐ Regular well-care visits (check-ups)
It is important to have yearly well-care check-ups. These visits will assist in checking your child’s health, giving shots, and answering questions about your child’s health.

☐ Monitor growth
It is important to check growth at every visit. Measurements include height, weight, and body mass index (BMI). These measurements are very important to assessing the overall health of the child. Discuss diet, activity level, and growth. Your child’s doctor can help with question about any need for vitamins or supplements.

☐ Immunizations (shots)
Your child’s doctor should follow the same shot schedule as for any other child. This includes yearly influenza (flu) shots. It may include other shots, too, depending on your child’s health history.

☐ Heart
The need to see a cardiologist (heart doctor) during this age is based on a child’s health history and exam. Let your child’s doctor know if your child is having increasing fatigue (low energy) or shortness of breath at rest or during activity. If new symptoms appear, an echocardiogram (an ultrasound picture of the heart) may be needed.

☐ Hearing
Hearing testing is recommended at least every year. This may be needed more often if your child has hearing or ear problems.

☐ Vision
Ophthalmology examination by an eye doctor is recommended at least every 3 years or more often if there are known eye or vision issues.

☐ Thyroid
The thyroid gland is usually normal in babies with Down syndrome. It can stop working normally for half of people with Down syndrome by adulthood. The symptoms of low thyroid can be hard to notice in Down syndrome, so a blood test (TSH) is needed every year, or sooner if symptoms change. When there is a problem, treatment is safe and can often be started by your primary doctor.

☐ Blood tests
Tests for low iron or anemia (hemoglobin and other tests if needed) should be done every year.

☐ Stomach or bowel problems (diarrhea, constipation)
Discuss toilet patterns at each visit, especially any ongoing problems with loose stools or constipation. These are common in children with Down syndrome. Some children with Down syndrome have celiac disease, which is a problem with tolerating some grains, including wheat. Testing can help to identify that condition, and may lead to changes in diet. Celiac disease can affect growth, stooling patterns, and behavior. Let your child’s doctor know if your child is having:

- Very loose stools
- Hard to treat constipation (hard or painful stools)
- Slow growth/weight loss
- Belly pain or stomach swelling
- New or challenging behavior problems

☐ Neck instability
Bones in the neck or spine can be unstable in some people with Down syndrome. There are almost always visible signs when there are problems. Daily physical activity is important to your child and should not be limited by unneeded worries. X-rays are not needed unless there is pain or changes in function. If x-rays are done, and the results are abnormal, your child may be referred to a spine or neck specialist. Special neck positioning may be needed for some medical procedures. Let your child’s doctor know if your child is having:

- Stiff or sore neck
- Change in stooling or urination
- Change in use of arms or legs
- Head tilt
- Change in walking
- Numbness (loss of normal feeling) or tingling in arms or legs
Sleep issues
Obstructive sleep apnea is a common problem for people with Down syndrome. This is a sleep problem that can affect a child’s behavior and ability to pay attention. It can also affect the heart. Some symptoms are obvious (snoring, restless sleep, waking at night, daytime sleepiness), but it can be hard to tell just by watching. AAP guidelines recommend that every child with Down syndrome have a sleep study by the age of 4 years. (That testing may be hard to find in some parts of the country.) Let your child’s doctor know if your child is having:

- Loud breathing
- Snoring
- Waking up often at night
- Daytime sleepiness
- Restless sleep (moving around a lot)
- Uncommon sleep positions (like sitting up to sleep or sleeping with neck arched back)
- Pauses in breathing during sleep
- Behavior problems

Skin
Discuss with your child’s doctor if your child has very dry skin or other skin problems.

Brain and nervous system
Discuss with your child’s doctor concerns about neurologic problems, such as seizures.

Behavior and mental health
Some youth with Down syndrome can have behavior or mental health issues that affect how they play and work at home, at school, or in the community. Let your child’s doctor know if your child is having problems that make it hard for him or her to function in the home, community, or school, including:

- Anxiety
- High activity level
- Wandering off
- Not listening
- Other behavior concerns
- Attention problems
- Sad mood or lack of interest in activities
- Loss of skills (not being able to do the things they used to do)
- Obsessive compulsive behaviors (behaviors that are repeated frequently)

Sexuality
Young people need help to learn the right kind of touch in social situations and the correct names for body parts. Puberty brings many changes and it may help to talk with your child’s doctor about several things. Discussions may include:

- Changes in puberty
- Managing sexual behaviors (such as masturbation)
- Gynecologic (women’s health) care for girls with Down syndrome, including periods and mood changes related to (menstrual) periods.
- Fertility, birth control, prevention of sexually transmitted infections
- Risk for a person with Down syndrome having a child with Down syndrome

Transitions
Discussion topics may include:

- School placement and goals for education. These should plan for transition and vocational (job skills) training.
- Guardianship and long-term financial planning.
- Adult work and places to live: family relationships, group homes and independent living opportunities, workshop settings, and other community-supported employment
- Working toward independence with bathing, grooming, self-care, and skills of community living
- Places for health care as an adult

Aging
Discussion of special health problems for adults with Down syndrome. This includes a tendency to early-aging and higher risk for Alzheimer disease in some people.