CLINICAL REPORT Guidance for the Clinician in Rendering Pediatric Care

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN™

# Health Supervision for Children and Adolescents With Down Syndrome

Marilyn J. Bull, MD, FAAP,[a] Tracy Trotter, MD, FAAP,[b] Stephanie L. Santoro, MD, FAAP,[c] Celanie Christensen, MD, MS, FAAP,[a] Randall W. Grout, MD, MS, FAAP,[d] THE COUNCIL ON GENETICS

This clinical report is designed to assist the pediatrician in caring for the child, adolescent, and family in whom a diagnosis of Down syndrome has been confirmed by chromosome analysis or suspected by prenatal screening. Although a pediatrician's initial contact with the child is usually during infancy, occasionally the pregnant woman who has been given a prenatal diagnosis of Down syndrome will be referred for review of the condition and genetic counseling; this report offers guidance for this situation, as well. Age-specific guidance for the clinician is provided in Supplemental Fig 1.

Pediatricians play an important role in the care of children and adolescents with Down syndrome and their families. Down syndrome is the most common chromosomal cause of intellectual disability, and there has been a significant improvement in quality of life for affected people. Awareness of the issues important to affected children, adolescents, and their caregivers can make a great difference in outcomes across the lifespan.

Children with Down syndrome may have many cooccurring medical conditions and cognitive impairment because of the presence of extra genetic material from chromosome 21 (Table 1).[1,2] Although the phenotype is variable, there typically are multiple features that enable the experienced clinician to suspect the diagnosis. Among the more common physical findings are hypotonia, small brachycephalic head, epicanthal folds, flat nasal bridge, upward-slanting palpebral fissures, Brushfield spots, small mouth, small ears, excessive skin at the nape of the neck, single transverse palmar crease, short fifth finger with clinodactyly, and wide spacing between the first and second toes, often with a deep plantar groove. The degree of cognitive impairment is variable and may be mild (IQ of 50–70), usually is moderate (IQ of 35–50), or occasionally is severe (IQ of 20–35).

Medical conditions common in children with Down syndrome include hearing loss (75%), obstructive sleep apnea (50%–79%), otitis media

[a]*Department of Pediatrics, Division of Developmental Medicine, Indiana University School of Medicine, Riley Hospital for Children, Indianapolis, Indiana;* [b]*San Ramon Valley Primary Care Medical Group, San Ramon, California;* [c]*Department of Pediatrics, Division of Medical Genetics and Metabolism, Massachusetts General Hospital, Boston, Massachusetts; and* [d]*Division of Children's Health Services Research, Department of Pediatrics, Indiana University School of Medicine, Indianapolis, Indiana*

Drs Bull, Trotter, Santoro, Christensen, and Grout were directly involved in the planning, researching, and writing of this report, approved the final manuscript as submitted, and agree to be accountable for all aspects of the work.



**DOI:** https://doi.org/10.1542/peds.2022-057010

Address correspondence to Marilyn J. Bull, MD, FAAP. E-mail: mbull@iu.edu

PEDIATRICS (ISSN Numbers: Print, 0031-4005; Online, 1098-4275).



**To cite:** Bull MJ, Trotter T, Santoro SL, et al; AAP Council on Genetics. Health Supervision for Children and Adolescents With Down Syndrome. *Pediatrics*. 2022;149(5):e2022057010

**TABLE 1** Medical Problems Common in Down Syndrome

| Condition | % |
|---|---|
| Hearing problems | 75 |
| Vision problems | 60–80 |
| Nystagmus | 3–33 |
| Glaucoma | <1–7 |
| Nasolacrimal duct occlusion | 3–36 |
| Cataracts | 3 |
| Strabismus | 36 |
| Refractive errors | 36–80 |
| Keratoconus | 1–13 |
| Obstructive sleep apnea | 50–79 |
| Otitis media with effusion | 50–70 |
| Congenital heart disease | 40–50 |
| Feeding difficulty | 31–80 |
| Respiratory infection | 20–36 |
| Dermatologic problems | 56 |
| Hypodontia and delayed dental eruption | 23 |
| Congenital hypothyroidism | 2–7 |
| Antithyroid antibody positive (Hashimoto thyroiditis; incidence dependent on age) | 13–39 |
| Hyperthyroidism | 0.65–3 |
| Thyroid disease by adulthood | 50 |
| Gastrointestinal atresias | 12 |
| Seizures | 1–13 |
| Hematologic problems | |
| Anemia | 1.2 |
| Iron deficiency | 6.7 |
| Transient abnormal myelopoiesis | 10 |
| Leukemia | 1 |
| Autoimmune conditions | |
| Hashimoto thyroiditis | 13–39 |
| Graves' disease | 1 |
| Celiac disease | 1–5 |
| Type 1 diabetes | 1 |
| Juvenile idiopathic arthritis | <1 |
| Alopecia | 5 |
| Symptomatic atlantoaxial instability | 1–2 |
| Autism | 7–19 |
| Hirschsprung disease | <1 |
| Moyamoya disease | Down syndrome 26 times greater in patients with Moyamoya than Down syndrome in live births |

(50%–70%), eye problems (60%–80%), including cataracts (<1%–3%),[3] nasolacrimal duct obstruction (3%–36%), and strabismus and severe refractive errors (36%–80%), congenital heart defects (50%), neurologic dysfunction (1%–13%), gastrointestinal atresia (12%), hip dislocation and hip abnormalities (2%–8%),[4,5] symptomatic atlantoaxial instability (1%–2%),[6,7] thyroid disease (24%–50%)[2,8] and, less commonly, transient abnormal myelopoiesis (4%–10%) and later leukemia (1%), autoimmune diseases,[2,8–10] including Hashimoto thyroiditis (13%–39%), with incidence dependent on age, celiac disease (1%–5%), Hirschsprung disease (<1%),[3] and autism (7%–19%).[11,12]

People with Down syndrome often function more effectively in social situations than would be predicted based on cognitive assessment results, unless there is presence of cooccurring autism. Although the level of social–emotional functioning may vary, these skills may be improved with early intervention and therapy through early adulthood.

In ~96% of children with Down syndrome, the condition is sporadic because of nonfamilial trisomy 21, in which there are 47 chromosomes with the presence of a free extra chromosome 21. In ~3% to 4% of people with the Down syndrome phenotype, the extra chromosomal material is the result of an unbalanced translocation between chromosome 21 and another acrocentric chromosome, usually chromosome 14 or 21. Approximately three-quarters of these unbalanced translocations are de novo, and the remainder result from translocation inherited from a parent. If the child has a translocation, the parents should be offered a karyotype to determine whether the translocation is familial or de novo. In the remaining 1% to 2% of people with the Down syndrome phenotype, a mix of 2 cell lines is present: 1 normal and the other with trisomy 21. This condition is called mosaicism. People with mosaicism may be more mildly affected than people with complete trisomy 21 or translocation chromosome 21, but this is not always the case, and their condition may include any of the associated medical problems and may be indistinguishable from trisomy 21. The chance of recurrence for families with an affected child depends on many factors and vary greatly, from 1% in most families to 100% in some circumstances. Table 2 describes the different chromosomal characteristics of Down syndrome.

Formal counseling by a clinical geneticist or genetic counselor is recommended for all families.

Several areas require ongoing assessment throughout childhood and should be reviewed at every

**TABLE 2** Chromosomal Basis of Down Syndrome

| Percentage | Chromosomal Basis |
|---|---|
| 96 | Meiotic nondisjunction (95% occur in egg, with recurrence risk of 1% until mother's age risk exceeds 1% at age 40, and it then increases according to maternal age) |
| 3–4 | Translocation (usually occurs with 1 chromosome 21 attached to chromosome 14, 21, or 22) |
| | 14/21 translocation (1/3 of patients have a parent carrier with balanced karyotype) |
| | 90% have mother as the carrier parent, with a recurrence chance of 10%–15% |
| | 10% have father as the carrier, with a recurrence chance of 2%–5% |
| | 21/21 translocation (1/14 of patients have parent carrier with a balanced karyotype); carrier parent equally likely mother or father, with recurrence chance of 100%[13] |
| 1–2 | Mosaicism: number of affected cells vary between individuals; clinical findings vary widely |
| | Medical complications fewer and intellectual disability often less severe |
| Partial trisomy: duplication of delimited segment of chromosome 21 present; extremely rare | |

Adapted from Bull MJ. Down syndrome.[14]
Information regarding meiotic nondisjunction and translocation is from Hook,[13] information regarding mosaicism is from Papavassiliou et al,[15] and information regarding partial trisomy is from Pelleri et al.[16]

health supervision visit and at least annually. These areas include:

- personal support available to family;
- participation in a family-centered medical home;
- age-specific Down syndrome-related medical and developmental conditions;
- financial and medical support programs and long-term financial planning for which the child and family may be eligible;
- injury and abuse prevention, with special consideration of developmental skills and intellectual ability; and
- nutrition and activity to maintain appropriate weight.

## THE PRENATAL VISIT

The American College of Obstetricians and Gynecologists recommends that all pregnant women, regardless of age or risk status, be offered the option of screening and diagnostic testing for Down syndrome.[17,18]

A wide variety of screening test options exist in the first and second trimester using maternal serum and ultrasonography. Each offers varying levels of sensitivity and specificity. No 1 screening test is superior to other screening tests in all characteristics. In recent years, noninvasive prenatal testing by cell-free DNA (cfDNA) has become available and is the most sensitive method for screening for Down syndrome. cfDNA screening for Down syndrome is significantly more sensitive and specific than conventional screening methods, with a 2017 meta-analysis reporting a detection rate of 99.7%, with a false-positive rate of 0.04% in singleton pregnancies.[19] cfDNA uses a maternal blood sample to analyze free-floating small fragments of DNA from the placenta. Because cfDNA is from the placenta and not directly from the fetus, it is a screening test and not diagnostic. cfDNA analysis can be performed as early as 9 to 10 weeks' gestation depending on the laboratory, and a high-risk result from cfDNA would require confirmation by diagnostic testing with chorionic villus sampling (CVS) or amniocentesis. Screening for trisomy 21 by cfDNA in twin pregnancies can be performed, but total number of reported cases is small.[20]

Other screening tests for Down syndrome include first-trimester screening, which incorporates maternal age, nuchal translucency ultrasonography, and measurement of maternal serum $\beta$ human chorionic gonadotropin and pregnancy-associated plasma protein A. Second-trimester screening is available for patients who first seek medical care in the second trimester or in locations where first-trimester screening is not available. The second-trimester serum screening, often called the quad screen, incorporates maternal age risk with measurement of maternal serum $\beta$ human chorionic gonadotropin, unconjugated estriol, $\alpha$-fetoprotein, and inhibin concentrations. The detection rate of Down syndrome by first-trimester screening is 82% to 87%, by second trimester screening is 80%, and by combined first- and second-trimester screening (referred to as integrated screening) is $\sim$95%. These screening tests are reported to have a 5% false-positive rate.[21–24]

Ultrasonography is an additional screening tool for Down syndrome because structural changes, including congenital heart defects, increased nuchal skin fold, "double bubble" sign suggestive of duodenal atresia, ventriculomegaly, and short–long bones, may be identified by prenatal imaging. Although ultrasonography is an additional screening tool, it is not diagnostic for Down syndrome.

Diagnostic testing for Down syndrome includes CVS or amniocentesis. CVS has the benefit of being performed earlier in pregnancy, between 10 and 14 completed weeks' gestation. A placental sample is obtained either transabdominally or transcervically, depending on provider preference and placental location. Amniocentesis is a transabdominal procedure to remove a sample of amniotic fluid performed after 15 weeks' gestational age. Risk for

procedure-related loss of pregnancy from CVS or amniocentesis is comparable in recent studies when performed by providers with expertise, 0.22% for amniocentesis and 0.11% for CVS.[25,26]

Pediatricians may be asked to counsel a family whose fetus has been identified with or is at increased chance of having Down syndrome. Families may have a great number of questions during any pregnancy and especially when the child will have Down syndrome. They may have received counseling from a certified genetic counselor, a clinical geneticist, maternal–fetal medicine specialist, obstetrician, or developmental specialist. In addition, parents may have received information and support from a family-led organization such as Parent to Parent USA, a local Down syndrome group, a national Down syndrome organization, social media, or possibly an Internet site with inaccurate information. Pediatricians who often have a previous relationship with the family may be the natural source of support for and guidance in the context of the medical home. The clinician should be prepared to respond to questions, review information the family has received, and assist in the decision-making process.[27] When asked, the pediatrician should discuss the following topics with the family:

1. The prenatal laboratory studies and any confirmatory testing that led to the diagnosis and any fetal imaging studies that have been or will be performed. Many families find it important to have the diagnosis confirmed before they can consider what it will mean to their infant and their family.
2. Families benefit from hearing a fair and balanced perspective, including the many positive outcomes of children with Down syndrome and their effects on the family. Families usually have questions about prognosis and phenotypic manifestations, including the wide range of variability seen in infants and children with Down syndrome. The prenatal visit is a good time to offer a connection to a peer-to-peer organization for support (see Family Resources).
3. Discuss any additional studies performed that may refine the estimation of the prognosis (eg, fetal echocardiography, ultrasonography for gastrointestinal tract malformations). Consultation with an appropriate medical subspecialist, such as a pediatric cardiologist or a pediatric surgeon, may occur prenatally if abnormal findings are detected.
4. Discuss currently available treatments and interventions. Families need to hear that they are not alone and that there are supports and services for them after the infant is born. Discuss early intervention resources, parent-support programs, and any appropriate future treatments.
5. Discuss extended life expectancy that has increased from 30 years in 1973 to 60 years in 2002. This increase has resulted from improved medical care, educational options, and enhanced social adaptation. Potential complications and adverse effects, costs and financial supports available, and other challenges associated with comprehensive management and care should also be discussed. The pediatrician can explain that they will be supported best in the context of a patient-centered medical home.
6. There are many issues for the family learning that their child will have Down syndrome to consider. These issues should be discussed using a nondirective approach. In cases of prenatal diagnosis, this may include discussion of pregnancy continuation or termination, raising the child in the family, foster care placement, and adoption.[28]
7. The mechanism for occurrence of Down syndrome in the fetus and the potential recurrence rate for the family, as provided by genetic counseling, should be shared. Discuss availability of genetic counseling or meeting with a genetics professional.

As the pregnancy continues, the pediatrician may:

1. Develop a plan for delivery and neonatal care with the obstetrician and the family. As the pregnancy progresses, additional studies should be performed if available, if recommended by medical subspecialists and/or if desired by the family. These studies (eg, detection of a complex heart defect by fetal echocardiography) may help direct development of a management plan and improve outcome for the mother and infant.[29]
2. Offer parent-to-parent contact and information about local and national support organizations because communication with experienced parents is often a very helpful resource for caregiver decision-making.
3. Offer referral to a clinical geneticist or genetic counselor for a more extended discussion of clinical outcomes and variability, recurrence rates, future reproductive options, and evaluation of the risks for other family members.

## HEALTH SUPERVISION FROM BIRTH TO 1 MONTH: NEWBORN INFANTS

It is recognized that the medical needs of newborn infants with Down syndrome vary, and the timing of each intervention depends on the infant's needs, but that it is important that all interventions are addressed and that careful transfer of care occurs at the time of discharge from the hospital.

### Examination

The first step in evaluating a newborn infant for trisomy 21 is a careful review of the family history and prenatal information, to include:

- results of prenatal chromosome studies, if performed; and
- family history of previous children born with trisomy 21 or developmental differences or pregnancies that ended in miscarriage. (These may be significant clues that a family may carry a balanced translocation that predisposes them to having children with trisomy 21.)

For children who have had the diagnosis made prenatally, a formal copy of the chromosome report from an amniocentesis or CVS should be obtained. This report allows the clinician to confirm the diagnosis, review the results with the family, and add the formal diagnosis to the child's medical record. If the results of prenatal testing are not available or if cfDNA alone was performed, a sample of cord or peripheral blood should be obtained for postnatal karyotype to confirm the diagnosis and rule out a chromosome translocation.

A physical examination is the most sensitive test in the first 24 hours of life to diagnose trisomy 21 in an infant. If the clinician believes that criteria for Down syndrome are present on physical examination, then a blood sample should be sent for a karyotype. The clinician should alert the laboratory and request rapid results. A study that uses fluorescent in situ hybridization (FISH) technology, in addition, should be available within 24 to 48 hours, if necessary, to facilitate diagnosis and parent counseling. A FISH study, however, can only indicate that an extra copy of chromosome 21 is present and does not determine the presence or absence of a translocation. Therefore, a positive FISH result should be confirmed by a karyotype to identify translocations that may have implications for further reproductive counseling for the parents and possibly other family members. A chromosomal microarray analysis is not appropriate because it will not differentiate trisomy 21 caused by nondisjunction versus an unbalanced translocation.

When delivering a diagnosis of Trisomy 21 (Table 3):

- the mother should be allowed to recover from the immediate delivery of the infant and have her partner or support person present before the diagnosis is given;
- the information should be relayed in a private setting by the physicians involved, optimally by the primary care provider for the infant and the delivering physician[30]; and
- it is recommended that hospitals coordinate the delivery of the information and offer a private hospital room pending confirmation of the diagnosis.

When providing information about Down syndrome to families, the physician should first congratulate parents on the birth of their infant. Obstetricians and pediatricians should coordinate their messaging and inform parents of their suspicion immediately, in a private setting, and when appropriate, with both parents together. Physicians should use their experience and

**TABLE 3** Communicating With Families[31]

| |
|---|
| At diagnosis, immediate advice remains pertinent regarding the need to: |
| • first, congratulate the family |
| • have infant present; refer to infant by name |
| • use a respectful bedside manner |
| • time discussion after labor is complete and as soon as diagnosis is suspected (not necessarily confirmed) |
| • have a support person present for mother, father, and family members as appropriate |
| • use a cohesive, physician-led team approach |
| Helpful discussion will include: |
| • up-to-date, accurate information |
| • a balanced approach rather than relying on personal opinions and experience |
| • person-first language (ie, child *with* Down syndrome)[32]; |
| • connection to other parents and resource groups |
| • discussion of life potentials for people with Down syndrome |
| Share with families the interplay within families and individual perspectives: |
| • individuals with Down syndrome: nearly 99% indicated that they were happy with their lives, and 97% liked who they are and encouraged health care professionals to value them, emphasizing that they share similar hopes and dreams as people without Down syndrome[33]; |
| • parents: 79% felt their outlook on life was more positive because of people with Down syndrome[31]; |
| • siblings: 88% felt that they were better people because of their siblings with Down syndrome[33]; |
| • a majority of families report unanimous feelings of love and pride |
| • positive themes dominate modern families[34] |

expertise in providing support and guidance for families. Clinicians should ensure a balanced approach that reflects the variability and broad range of current outcomes, rather than their personal opinions or experience, give current printed materials, and offer access to other families who have children with Down syndrome and support organizations if locally available. It is important that clinicians be cognizant of the realities and possibilities for people with Down syndrome to have healthy, productive lives.[30]

The laboratory diagnosis of Down syndrome should be confirmed, and the karyotype should be reviewed with the parents when the final result is available. The specific findings should be discussed with both parents whenever possible, including the potential clinical manifestations associated with the syndrome. These topics should be reviewed again at a subsequent meeting. Families should be offered referral for genetic counseling if it was not conducted prenatally.

Newborn care is often provided in a hospital setting by a physician who will not be the primary care pediatrician. If the physician providing newborn care will not be the primary care pediatrician, he or she should ensure that there is a smooth transition by transferring medical records and ensuring that an early newborn follow-up appointment is scheduled.

Characteristics attributable to Down syndrome, as well as those that are familial, should be discussed.

### Discuss and Review

- Hypotonia.
- Facial appearance and acknowledge the presence of familial characteristics.
- Cutis marmorata; explain that this is common in infants with Down syndrome and provide reassurance to family about this finding.

### Evaluate For

- Heart defects (~50% risk). Perform an echocardiogram, to be read by a pediatric cardiologist, regardless of whether a fetal echocardiogram was performed. Refer to a pediatric cardiologist for evaluation any infant whose postnatal echocardiogram results are abnormal.
- Feeding problems. Feeding difficulties including gastroesophageal reflux and dysphagia are extremely common (31%–80% in Down syndrome).[35,36] Dysphagia can result from both oromotor problems and oropharyngeal dysfunction. Hypotonia, relative macroglosia with a relatively small oral cavity, decreased jaw strength, and poor tongue control contribute to the problems. Symptoms of feeding difficulty include slow feeding, choking with feeds, and slow weight gain. Up to 90% of patients with Down syndrome who aspirate do so silently without cough or overt symptoms, and symptoms often are not recognized during a clinical feeding evaluation.[35,37] Feeding difficulty occurs with increased frequency in all infants with Down syndrome, but especially those who are born preterm, have marked hypotonia, are underweight, or have desaturation with feeds. Infants who (1) have marked hypotonia as judged by the pediatrician, (2) are underweight, (3) have slow feeds, (4) have choking with feeds, (5) have recurrent or persistent respiratory symptoms, or (6) desaturate with feeds should be referred promptly for skilled feeding assessment or possible video feeding study.[37,38] Video feeding studies can be helpful for determining which infants require intervention. Nonradiologic videofluoroscopic swallow studies, where available, may be performed for infants, including those who are breastfed. Feeding function changes over infancy and early childhood, and repeat studies may be indicated, especially if respiratory symptoms persist.[39] If untreated, aspiration is an overlooked cause of recurrent respiratory symptoms.[40] Infants with Down syndrome can breastfeed successfully, but some may need early support until a successful nursing pattern is established. Some infants may sleep for prolonged periods and if not gaining weight adequately, need to be awakened for feeds to maintain adequate calorie intake.
- Cataracts at birth by looking for a red reflex. Cataracts may progress slowly and, if detected, require prompt evaluation and treatment by an ophthalmologist with experience in managing the child with Down syndrome, because surgical outcomes in these cases are reassuring.[41,42]
- Congenital hearing loss, with objective testing, such as brainstem auditory evoked response or otoacoustic emission. If the infant did not pass newborn screening studies, refer to an otolaryngologist who is experienced in examining infants with stenotic external canals to determine whether a middle-ear abnormality is present. Tympanometry may be necessary if the tympanic membrane is poorly visualized.[42,43] Refer to early intervention within 48 hours of confirmation that the infant is deaf or hard of hearing.[43,44]
- Duodenal atresia or anorectal atresia/stenosis by obtaining a history and performing a clinical examination.
- Evaluation for apnea, bradycardia, or oxygen desaturation should occur with the infant in a

car safety seat, because all infants with Down syndrome are at increased risk attributable to hypotonia, upper airway obstruction, or having had cardiac surgery. A car safety seat screen should be conducted before hospital discharge.[45]

- Constipation. If constipation is present, evaluate for restricted diet or limited fluid intake, hypotonia, hypothyroidism, or gastrointestinal tract malformation, including stenosis or Hirschsprung disease, for which there is an increased risk. Review the timing of the passing of meconium because a delay may indicate Hirschsprung disease and other considerations.
- Gastroesophageal reflux, which is usually diagnosed and managed clinically. If contributing to cardiorespiratory problems or failure to thrive, refer for subspecialty intervention.
- Stridor, wheezing, or noisy breathing. If contributing to cardiorespiratory problems or feeding difficulty, refer to an otolaryngologist, pediatric pulmonologist, or aerodigestive program to assess for airway anomalies. Small nasal passages and nasal congestion often contribute to stridor. Tracheal anomalies and small tracheal size may also make intubation more difficult. Hypotonia and small tracheal size also increase the risk of recurrent episodes of croup.
- Hematologic abnormalities. Obtain a complete blood cell count with differential by 3 days of age to evaluate for transient abnormal myelopoiesis (TAM) (formerly called transient myeloproliferative disorder), polycythemia, and other hematologic abnormalities. Leukocytosis or TAM is relatively common in this population (9%) and can present with pericardial and pleural effusions, but can be silent without hepatosplenomegaly, jaundice, or rash.[46,47] Although leukocytosis or TAM usually regresses spontaneously within the first 3 months of life, infants with TAM may require chemotherapy and are at risk for death in the first 6 months of life (up to 20%), and have an increased risk of acute myeloid leukemia in the first 4 years of life (~30%). All infants with Down syndrome and TAM should be evaluated by pediatric hematology/oncology as soon as they are diagnosed. Numerous hematologic abnormalities other than >10% blasts are commonly reported in newborn infants with trisomy 21, including neutrophilia (80%), thrombocytopenia (66%), and thrombocytosis, which generally resolve in the first week of life, whereas macrocytosis is also common but often does not resolve.[48] Infants with numeric abnormalities other than macrocytosis that persist after the first week of life should be referred to a hematologist. Leukemia is more common in children with Down syndrome than in the general population but is still rare (1%).[49]
- Caregivers of infants with TAM should be counseled regarding the risk of leukemia and made aware of the signs, including easy bruising, recurring fevers, bone pain, easy bruising or bleeding, petechiae, onset of lethargy, or change in feeding patterns. Although leukemia is rare, children with Down syndrome are at increased risk to develop both acute lymphoblastic leukemia and acute myeloid leukemia, even without a history of TAM as a newborn infant.
- Polycythemia. Unrelated to congenital heart disease, polycythemia is common in the first week of life in Down syndrome (33%) and may persist for several months. Persistent polycythemia requires regular follow-up with blood counts until resolution.
- Congenital hypothyroidism (2%–7% risk).[8,50] Obtain thyroid-stimulating hormone (TSH) concentration if state's newborn screening measures only free thyroxine (T4); congenital hypothyroidism can be missed if only the T4 concentration is obtained in the newborn screening. Many children with Down syndrome (25%–60%) have mildly elevated TSH and normal free T4 concentration (subclinical hypothyroidism), and hyperthyroidism occurs in 0.65% to 3%.[51] Elevated antithyroid antibodies occur frequently and, when present, increase the risk of later hypothyroidism.[50] By late childhood, the incidence of thyroid abnormality is 50%.[8,50] Management of children with abnormal TSH or T4 concentrations should be discussed with a pediatric endocrinologist.

### Anticipatory Guidance Given Between Birth and 1 Month of Age

- Discuss the strengths of the child and positive family experiences.
- Discuss the individual resources for support, such as family, religion, and friends.
- Talk about how and what to tell siblings, other family members, and friends. Review methods of coping with long-term disabilities (see "Resources for Families").
- Discuss efficacy of early intervention and availability of early intervention services and therapies in the community. Initiate referral for speech, fine motor, or gross motor therapies, unless medically contraindicated. Encourage families to participate in selection of therapies and therapists. Counsel families to share their impressions of their infant's strengths and progress with therapists and to actively participate in therapy sessions.

- Share information for local Down syndrome family and support groups, current books and pamphlets, and referrals for community and financial resources (see Resources for Families).
- Discuss increased susceptibility to respiratory tract infection. Children with signs and symptoms of lower respiratory tract infection should be evaluated acutely by a medical provider, and in the presence of cardiac or chronic respiratory disease, prompt diagnosis and treatment should be instituted.[40,52]
- Children with cooccurring conditions including qualifying congenital heart disease, airway clearance issues, or prematurity (born at <29 weeks, 0 days' gestation) may be considered for administration of respiratory syncytial virus prophylaxis.[53]
- Discuss with families the importance of cervical spine-positioning precautions to avoid excessive extension or flexion during any anesthetic, surgical, or radiographic procedure.
- Using the previously obtained karyotype, review the chance of recurrence in subsequent pregnancies and the availability of prenatal testing options, as discussed in previous genetic counseling.
- Discuss treatments that are considered complementary or alternative. Families need an opportunity to learn objectively which therapies are safe and which are potentially dangerous (eg, cell therapy that may transmit slow viruses and high doses of fat-soluble vitamins that can cause toxicity).
- Determine whether the child receives supplements, herbs, teas, or other treatments or supplements not previously discussed. Approximately 38% of parents of children with Down syndrome report using dietary supplements in their children, and 20% report they have not informed their pediatrician, usually because they have not been asked.[54] Several articles and websites provide useful information for clinicians and families.[54–55]
- Renal and urinary tract anomalies have been reported to occur at increased frequency among people with Down syndrome.[56] Although routine postnatal screening for renal anomalies is not currently recommended, if renal abnormalities are detected on prenatal ultrasonography, standard assessment would be required.

## HEALTH SUPERVISION FROM 1 MONTH TO 1 YEAR: INFANCY

### Physical Examination and Laboratory Studies

Follow *Bright Futures* schedule or more frequently if indicated.

- Obtain a history and perform a physical examination.
- Monitor weight and follow weight-for-length trends at each health care visit. Review the infant's growth and plot it on the Down syndrome-specific charts for weight, length, weight for length, and head circumference (available at www.cdc.gov).[57,58]
- Review feeding at every health supervision visit, ensure adequate iron intake, and inquire about any changes in respiratory symptoms with feeding (see "Health Supervision From Birth to 1 Month" for discussion).
- Review the previous hearing evaluation (brainstem auditory evoked response [BAER] or otoacoustic emission). If the infant passed the newborn screening study, rescreen at 6 months of age for confirmation.
- Risk of otitis media with effusion is 50% to 75%.[43] Middle-ear disease should be treated immediately when diagnosed. As soon as a clear ear is established, a diagnostic BAER should be performed to accurately establish hearing status.
- In children with stenotic canals in which the tympanic membranes cannot be seen, refer to an otolaryngologist as soon as possible for examination under an office microscope. Interval ear examinations should be performed by the otolaryngologist every 3 to 6 months until the tympanic membrane can be visualized by the pediatrician and tympanometry can be performed reliably.[43]
- A behavioral audiogram may be attempted at 1 year of age, but many children will not be able to complete the study. If unable to complete a behavioral audiogram, additional testing by BAER should be performed at 1 year.
- Ear anomalies also place child at risk for sensorineural hearing loss and vestibular problems that may affect balance, making the thorough audiologic assessment additionally important.[59–61]
- Within the first 6 months of life, refer to a pediatric ophthalmologist or ophthalmologist with expertise and experience with infants with disabilities to evaluate for strabismus, cataracts, nasolacrimal duct obstruction, refractive errors, glaucoma, and nystagmus.[16,62–64]
- Check the infant's vision at each visit and use developmentally appropriate subjective and objective criteria. If lacrimal duct obstruction is present, refer for evaluation for surgical repair of drainage system if not resolved by 9 to 12 months of age.[65]
- Verify results of newborn thyroid-function screen if not previously reviewed. Because of increased risk of acquired thyroid disease, repeat

measurement of TSH at 6 and 12 months of age and then annually (see Health Supervision From Birth to 1 Month for discussion).

- Monitor infants with cardiac defects at all well-child visits, typically ventricular or atrioventricular septal defects that cause intracardiac left-to-right shunts, for symptoms and signs of congestive heart failure as pulmonary vascular resistance decreases and pulmonary blood flow increases. Tachypnea, feeding difficulties, and poor weight gain may indicate heart failure. Medical management, including nutritional support, may be required until the infant is in optimal condition to undergo cardiac surgery to repair the defects to limit the potential for development of pulmonary hypertension and associated complications.[66] Infants and children with Down syndrome are also at increased risk of pulmonary hypertension even in the absence of intracardiac structural defects. Close coordination of care between the primary care physician and the subspecialist is important for these infants.
- Anemia/iron deficiency: Obtain a complete blood cell count (CBC) with differential and either (1) a combination of ferritin and C-reactive protein (CRP), or (2) a combination of serum iron and total iron-binding capacity (TIBC), beginning at 1 year of age and annually thereafter.

Children with Down syndrome have been shown to have a similar risk for iron-deficiency anemia as the typical population, but it may be missed because of macrocytosis.[67] Iron insufficiency may precede iron-deficiency anemia and also can have long-term neurologic effects.[67,68] Macrocytosis, with increased erythrocyte mean corpuscular volume, is present in up to one-third of patients with Down syndrome.[69] Thus, a low mean corpuscular volume is not a useful screen for the diagnoses of iron deficiency/insufficiency, lead toxicity, or thalassemia in children with Down syndrome. Screening by hemoglobin concentration identifies iron deficiency anemia but misses iron deficiency/iron insufficiency. Using the CBC parameter of an elevated relative distribution width with ferritin or transferrin saturation or serum iron divided by TIBC leads to 100% sensitivity in identifying iron insufficiency, iron deficiency, or anemia. Serum ferritin concentration is an acute-phase reactant and is not useful if inflammation is present or CRP is elevated: subsequent evaluation with iron concentration and TIBC may be needed to confirm diagnosis.

- Although not unique to children with Down syndrome, low ferritin is also associated with sleep problems, and iron deficiency may be considered in differentials for children with sleep difficulty.[70] A physician may prescribe iron supplementation for children with sleep problems and a ferritin concentration $<50$ μg/L.[70,71]
- Pediatricians should be alert to the signs and symptoms of leukemia discussed in Health Supervision From Birth to 1 Month and obtain an extra CBC with differential if symptoms occur. Children with Down syndrome who develop acute leukemia can be treated successfully with similar acute lymphocytic leukemia therapy or de-intensified acute myeloid leukemia chemotherapy regimens with outcomes superior to other children.[72,73]
- Assess with complete neurologic history and examination and consult with neurology as needed for signs of neurologic dysfunction that may occur. Children with Down syndrome have an increased risk of seizures, including infantile spasms (1%–13%)[74,75] and other conditions, including moyamoya disease[76,77] and benign movement disorders such as shuddering.
- Administer immunizations, including influenza vaccine, respiratory syncytial virus vaccine for infants with cooccurring qualifying conditions, and other vaccines recommended for all children, unless there are specific contraindications.[78]
- Assess for dermatologic findings and advise parents that xerosis (dry skin) and cutis marmorata are common.
- At least once during the first 6 months of life, discuss with family symptoms of obstructive sleep apnea, including heavy breathing, snoring, uncommon sleep positions, frequent night awakening, daytime sleepiness, apneic pauses, and behavior problems that could be associated with poor sleep. Refer to a physician with expertise in pediatric sleep disorders for examination and further evaluation of a possible sleep disorder if any of the previously mentioned symptoms occur.[79,80]
- At each well-child visit, discuss with parents the importance of maintaining the cervical spine in a neutral position during any anesthetic, surgical, or radiographic procedure to minimize the risk of spinal cord injury and review the signs and symptoms of myelopathy, which include asymmetry of movement, weakness, and, on examination, increased deep tendon reflexes. Obtain history and carefully perform a physical examination, paying attention for myelopathic signs and symptoms.

### Anticipatory Guidance From 1 Month to 1 Year

- Review availability of resources, including Down syndrome support groups and organizations that help with navigation of community and financial resources, at least once in the first year of life (see Resources for Families).
- Assess the emotional status of caregivers and intrafamilial relationships at each well-child visit. Share information for support, including respite care and caregiver counseling, as desired. Inquire about how siblings are adjusting to the new baby and offer education to support the siblings as needed.
- Review connection to early intervention services and their relationship to the strengths and needs of the infant and family at each well-child visit. Ensure that the family knows how to implement early intervention therapy recommendations on a daily basis.
- Review the family's understanding of the chance of recurrence of Down syndrome and the availability of prenatal diagnosis and/or screening at least once in the first year of life, and more often if judged necessary by the clinician. Offer referral for genetic counseling if desired by the family.
- Be prepared to discuss and answer questions about treatments that are considered complementary and alternative at each well-child visit.

## HEALTH SUPERVISION FROM 1 TO 5 YEARS: EARLY CHILDHOOD

Follow *Bright Futures* schedule or more frequently if indicated.

- Obtain a history and perform a physical examination.
- Monitor weight and follow weight-for-height trends at each health care visit. Review the infant's growth and plot it on the Down syndrome-specific charts for weight, length, weight for length <2 years of age, BMI for age 2 to 10 years, and head circumference (available at www.cdc.gov).[58]
- Ask about changes in feeding or any changes in respiratory symptoms with feeding and ensure adequate iron intake (see Health Supervision From Birth to 1 Month for discussion).
- Anemia/iron deficiency: Obtain a CBC with differential and either (1) a combination of ferritin and CRP, or (2) a combination of serum iron and TIBC, beginning at 1 year of age and annually thereafter[69] (see "Health Supervision From 1 Month to 1 Year" for discussion).
- Low ferritin is also associated with sleep problems, particularly restless leg syndrome, and iron deficiency may be considered in the differential diagnosis for children with sleep difficulty.[70] A physician may prescribe iron supplementation for children with restless sleep and a ferritin concentration <50 μg/L.[70,71]
- Solid tumors: In contrast to the increased risk of leukemia, compared with the general population, the overall risk for solid tumors is not increased in Down syndrome. Although rare, solid tumors may occur, and clinicians should remain alert to this possibility.[81] Some are very rare (breast cancers, neuroblastoma, and medulloblastoma), and some do not differ significantly from the general population (gastric, colon, and ovarian cancers and gliomas).[82] Importantly, testicular cancer is the only solid tumor that is more common in Down syndrome.[83] Clinicians should palpate the testes during routine health supervision examinations for any changes, including development of a lump or swelling. Patients with Down syndrome may not recognize testicular changes that could be a sign of testicular cancer. Although there is not clear evidence that screening is beneficial, the physician may recommend routine screening for testicular cancer by a trusted adult.[84]
- Review the risk of hearing loss associated with otitis media with effusion.
  - For a child who passed diagnostic hearing testing, behavioral audiogram and tympanometry should be performed every 6 months until normal hearing levels are established bilaterally by ear-specific testing (usually after 4 years of age).
  - Subsequently, behavioral hearing tests should be performed annually. If normal hearing is not established by behavioral testing, additional screening by otoacoustic emissions or diagnostic BAER should be performed, with sedation if necessary.
  - Children who demonstrate hearing loss should be referred to an otolaryngologist who is comfortable with the examination of children with stenotic ear canals. The risk of otitis media with effusion between 3 and 5 years of age is ~50% to 70%. If middle ear disease occurs, obtain developmentally appropriate hearing evaluation after treatment.
  - Discuss with caregivers the importance of optimal hearing for speech development and learning.
- Check the child's vision, and use developmentally appropriate subjective and objective criteria, including photoscreening if available, at each well-child visit.[85] Refer the child with abnormal findings on photoscreening or annually if photoscreening is not available to a pediatric

ophthalmologist or ophthalmologist with special expertise and experience with children with disabilities. Children with Down syndrome have a 50% risk of refractive errors that lead to amblyopia between 3 and 5 years of age. Addressing refractive errors and strabismus at an early age can help prevent amblyopia and encourage normal visual development.[63,85,86]

- Atlantoaxial instability: Discuss with parents, at least biennially, the importance of cervical spine-positioning precautions for protection of the cervical spine during any anesthetic, surgical, or radiographic procedure. Perform careful history and physical examination with attention to myelopathic signs and symptoms at every well-child visit or when symptoms possibly attributable to spinal cord impingement are reported. Parents should also be instructed to contact their physician for new onset of symptoms of change in gait or use of arms or hands, change in bowel or bladder function, neck pain, stiff neck, head tilt, torticollis, how the child positions his or her head, change in general function, or weakness.

### The Child Without Symptoms of Atlantoaxial Instability

- Children with Down syndrome are at slightly increased risk of symptomatic atlantoaxial subluxation.[6,7] However, the child <3 years does not have adequate vertebral mineralization and epiphyseal development for accurate radiographic evaluation of the cervical spine.[87] Plain radiographs do not predict well which children are at increased risk of developing spine problems, and normal radiographs do not provide assurance that a child will not develop spine problems later.[88,89] For these reasons, routine radiologic evaluation of the cervical spine in asymptomatic children is not recommended. Current evidence does not support performing routine screening radiographs for assessment of potential atlantoaxial instability in asymptomatic children.[7,90–93]
- Special Olympics requires documentation of physical examination of all athletes for participation in sports.[94]

### The Child With Symptoms of Possible Atlantoaxial Instability

- Any child who has significant neck pain, radicular pain, weakness, spasticity or change in tone, gait difficulties, hyperreflexia, change in bowel or bladder function, or other signs or symptoms of myelopathy must undergo plain cervical spine radiography in the neutral position.[95] If significant radiographic abnormalities are present in the neutral position, no further radiographs should be taken and the patient should be referred as quickly as possible to a pediatric neurosurgeon or pediatric orthopedic surgeon with expertise in evaluating and treating atlantoaxial instability. If no significant radiographic abnormalities are present, flexion and extension radiographs may be obtained in collaboration with the subspecialist before the patient is promptly referred.[92,93,95]
- Discuss with caregivers that trampoline use should be avoided by all children, with or without Down syndrome, unless part of a structured training program with appropriate supervision and safety measures in place.[96] Parents can be advised that participation in contact sports, such as football, soccer, and gymnastics, places children at risk for spinal cord injury.[97]
- Measure TSH annually or sooner if child has symptoms that could be related to thyroid dysfunction (see Health Supervision From Birth to 1 Month for discussion). Measure TSH every 6 months if antithyroid antibodies were previously detected.
- For children on a diet that contains gluten, review for symptoms potentially related to celiac disease at each health supervision visit because children with Down syndrome are at increased risk. These symptoms include diarrhea or protracted constipation, slow growth, unexplained failure to thrive, anemia, abdominal pain or bloating, or refractory developmental or behavioral problems.[97–99] For those with symptoms, obtain a tissue transglutaminase immunoglobulin A (TTG IgA) concentration and simultaneous quantitative IgA. The quantitative IgA is important, because an IgA deficiency renders the TTG IgA unreliable. Refer patients with abnormal laboratory values for specialty assessment. Do not institute a gluten-free diet before confirmation of the diagnosis, because lack of gluten can make interpretation of endoscopic results difficult. There is no evidence that routine screening of asymptomatic individuals would be beneficial. There are neither data nor consensus that would indicate whether patients with persistent symptoms who had normal laboratory values on initial evaluation should have further laboratory tests.
- Discuss symptoms of sleep-disordered breathing, including heavy breathing, snoring, restless sleep, uncommon sleep positions, frequent night awakening, daytime sleepiness, apneic pauses, and behavior problems that could be associated with poor sleep at each well-child visit. There is poor correlation between negative parent-report of symptoms and polysomnogram results.[80,100]

Therefore, referral to a pediatric sleep laboratory for a sleep study or polysomnogram for all children with Down syndrome between ages 3 and 4 years is recommended. Refer to a physician with expertise in pediatric sleep any child with signs or symptoms of obstructive sleep apnea or abnormal sleep-study results. Children who have adenotonsillectomy for treatment of obstructive sleep apnea should have a repeat polysomnogram after surgical intervention, because there is significant incidence of persistent air obstruction that requires additional evaluation and intervention.[101,102] It is recognized that access to a pediatric sleep laboratory or specialist may be limited for some populations and geographic areas.

- Discuss obesity as a risk factor for sleep apnea.[103]
- Recognize that sleep disturbance is extremely distressful to families. Low ferritin is also associated with sleep problems, particularly restless leg syndrome, and iron deficiency may be considered in the differential diagnosis for children with sleep difficulty.[70] A physician may elect to prescribe iron supplementation for children with restless sleep and a ferritin concentration $<50$ μg/L.[70,71]
- Remind the family to maintain follow-up with a pediatric cardiologist, per specialist recommendation, for patients with cardiac lesions, even after complete repair to monitor for recurrent/residual lesions, as well as possible development of pulmonary hypertension.
- Discuss with caregivers at every health supervision visit the child's behavioral and social progress. Encourage families to teach self-help skills and counsel to prevent wandering. Refer children who may have autism spectrum disorder, attention-deficit/hyperactivity disorder, or other psychiatric or behavioral problems for appropriate evaluation and intervention as soon as suspected.[104] Autism and other behavioral problems occur with increased frequency in children with Down syndrome, and symptoms may manifest as early as 2 or 3 years of age.[11,12,105–107]
  - A variety of screening tools have been used to identify children who may have a dual diagnosis of Down syndrome with autism spectrum disorder, although none have been studied in a large population. Examples (not an exhaustive list) include the Childhood Autism Rating Scale, the Social Communication Questionnaire, the Aberrant Behavior Checklist, and the Autism Behavior Checklist.[11,12,108,109]
- The diagnosis of autism spectrum disorder in children with Down syndrome is often delayed, because presentation can be subtly different from children with idiopathic autism spectrum disorder. Children with Down syndrome and autism spectrum disorder have better imitation, relating, and receptive skills when compared with children with autism spectrum disorder without Down syndrome. However, these adaptive skills are impaired in children with a dual diagnosis when compared with children with Down syndrome alone.[110] Also, when compared with children with Down syndrome alone, children with dual diagnosis exhibit more stereotypies, repetitive language, overactivity, social withdrawal, anxiety, and self-injury.[12,109] There is also decreased receptive and expressive language skills, as well as cognitive skills in children with a dual diagnosis.[105] Given these differences, specialty evaluation is needed to make an appropriate diagnosis of autism spectrum disorder in children with Down syndrome. The pediatrician should screen all children with Down syndrome for autism, as they would other children, between 18 and 24 months of age, and refer those with a concerning screen for specialty evaluation.[111] It is important to avoid assuming symptoms of autism are the known delays related to Down syndrome, referred to as overshadowing. Referral as soon as an autism diagnosis is suspected is critical, because early treatment is important in all children with autism spectrum disorder, including those with Down syndrome.
- Inquire about symptoms of neurologic dysfunction, including seizures, and perform a neurologic examination. Pediatricians should be aware of symptoms referred to as "acute regression in Down syndrome," "catatonia," or "disintegrative disorder" occurring in late childhood, adolescence, or early adulthood. Patients who experience loss of skills, marked mood changes, or catatonia, or who develop repetitive thoughts or behaviors that interfere with usual life activity, should be referred to specialists familiar with diagnosis and treatment of the disorder.[112,113]
- Skin problems are particularly common in patients with Down syndrome. Xerosis (very dry skin) or hair thinning may be a sign of hypothyroidism and warrant an interim TSH. Be attentive to dermatologic issues that may have an autoimmune etiology and are prevalent among children with Down syndrome, such as alopecia areata and vitiligo.

Folliculitis and keratosis pilaris are also commonly seen in children with Down syndrome. Assess for skin findings, discuss them with the patient and family, and consider referral to a dermatologist if needed.[114,115]

### Anticipatory Guidance From 1 to 5 Years

- Review early intervention, including physical therapy, occupational therapy, and speech therapy, at all health supervision visits.
- Discuss at the 30-month visit the transition from early intervention to preschool, which occurs at 36 months of age. Help the family understand the change from the Individualized Family Service Plan in early intervention to the Individualized Education Program through public education (see Resources for Families).
- Review availability of resources, including Down syndrome support groups and organizations that help with navigation of community and financial resources, including child care (see Resources for Families).
- Provide influenza vaccine annually. Respiratory syncytial virus prophylaxis may be considered for children <2 years who have cooccurring qualifying conditions. Children with chronic cardiac or pulmonary disease should be given the 23-valent pneumococcal polysaccharide vaccine at 2 years of age or older.[53]
- Reassure parents that delayed and irregular dental eruption patterns are common and that hypodontia occurs with increased frequency (23%).[116,117]
- Encourage and model use of accurate terms for genitalia and other private body parts (penis, vulva) anytime these body parts are discussed or examined. Model respect for body rights by reminding patients that their body is their own and explain what you will do before moving into their personal space or performing a procedure. Remind patient and family that the only reason anyone should be looking at or touching private body parts is for health (doctor office visits) or hygiene (bathing or showering).[118]
- On at least 1 well-child visit, educate the family about increased risk of sexual exploitation, and remind them that people their child knows and trusts are more likely than strangers to be perpetrators.
- At least once between 1 and 5 years of age, discuss future parental pregnancy planning and review chance of recurrence of Down syndrome and availability of prenatal testing options. Offer referral for genetic counseling if desired by the family.
- Assess the child's behavior and talk about behavioral management, sibling adjustments, socialization, and recreational skills.[119]
- Encourage families to establish optimal dietary and physical exercise patterns that will prevent obesity.
- Be prepared to discuss and answer questions about treatments that are considered complementary or alternative (see Health Supervision From Birth to 1 Month for discussion).

## HEALTH SUPERVISION FROM 5 TO 12 YEARS: LATE CHILDHOOD

Follow *Bright Futures* schedule or more frequently as indicated.

- Obtain a history and perform a physical examination.
- Monitor weight and follow BMI trends at each health care visit. Review the child's growth and plot it on the Down syndrome-specific charts for weight, height, and head circumference.[120] These charts should be used in conjunction with the Down syndrome-specific BMI chart for children up to age 10 and with the BMI chart from the Centers for Disease Control and Prevention, which is a better indicator of excess adiposity for children with Down syndrome over the age of 10.[58]
- Review feeding. Ask about any changes in respiratory symptoms with feeding and ensure adequate iron intake (see Health Supervision From Birth to 1 Month for discussion).
- Emphasize healthy diet and lifestyle for preventing obesity.
- Obtain annual ear-specific audiologic evaluation (see Health Supervision From 1 Month to 1 Year for discussion). If middle ear disease occurs, obtain developmentally appropriate hearing evaluation after treatment.
- Obtain ophthalmologic evaluation by photoscreening, if available, at every health supervision visit or by a pediatric ophthalmologist or ophthalmologist with expertise in children with disabilities every 2 years[63,85] (see Health Supervision From 1 Month to 1 Year for discussion).
- Measure TSH annually; the risk of hypothyroidism increases with age (See "Health Supervision From 1 to 5 Years" for discussion). Measure TSH every 6 months if antithyroid antibodies have been detected.
- Individualize cardiology follow-up on the basis of history of cardiac defects.
- Obtain a CBC and either (1) a combination of ferritin and CRP, or (2) a combination of serum iron and TIBC, beginning at 1 year of age and annually thereafter (see Health Supervision From 1 Month to 1 Year for discussion).
- A physician may prescribe iron supplementation for children with sleep problems and a ferritin concentration <50 μg/L (see Health Supervision From 1 to 5 Years for discussion).

- Palpate testes at each health supervision visit (see Health Supervision From 1 to 5 Years for discussion).
- For children on a diet that contains gluten, review for symptoms potentially related to celiac disease at every health maintenance visit and evaluate if indicated (see Health Supervision From 1 to 5 Years for discussion).
- At each well-child visit, discuss with family the importance of universal precautions for protection of the cervical spine during any anesthetic, surgical, or radiographic procedure. Perform careful history and physical examination, with attention to myelopathic signs and symptoms. Caregivers should also be instructed to contact their physician immediately for new onset of symptoms of myelopathy (see Health Supervision From 1 to 5 Years for discussion).
- Discuss skin, hair, and scalp care at each preventive health care visit and refer to dermatologist if needed (see Health Supervision From 1 to 5 Years for discussion).
- Encourage caregivers to promote self-help skills and assume developmentally appropriate responsibilities in the home. Monitor for behavior problems that interfere with function in the home, community, or school. Attention problems, attention-deficit/hyperactivity disorder, obsessive-compulsive behaviors, noncompliant behavior, and wandering off are some of the common behavior concerns reported. Psychiatric disorders affecting typically developing children may also occur. Evaluate for medical problems that can be associated with behavior changes, including thyroid abnormalities, celiac disease, sleep-disordered breathing, gastroesophageal reflux, and constipation. Intervention strategies depend on the child's age, the severity of the problem, and the setting in which the problem occurs. When symptoms interfere with daily activities, refer to community treatment programs, psychosocial services for consultative care, or behavioral specialists experienced in working with children with special needs. Refer patients who have chronic behavioral problems or manifest acute deterioration in function for specialized evaluation and intervention[112,113,121] (see Health Supervision From 1 to 5 Years for discussion).
- Be aware that children with Down syndrome are frequently more sensitive to certain medications. Before initiating medication for behavior management, the process should be discussed between the primary care physician and specialists involved in the child's care. Although there has been little research to directly address the use of psychotropic medications among children with Down syndrome, anecdotal reports indicate that these children may differ in their response to medications. Experience has led to the recommendation to start medications at the lowest recommended dose and increase or decrease the dose according to the child's response.[122]
- Inquire regarding symptoms of neurologic dysfunction, including seizures, and perform a neurologic examination.
- Discuss symptoms related to sleep-disordered breathing at every well-child visit, including snoring, restless sleep, daytime sleepiness, nighttime awakening, behavior problems, and abnormal sleep position. Refer to a physician with expertise in pediatric sleep, otolaryngologist, or a pediatric sleep medicine specialist any child with signs or symptoms of sleep-disordered breathing or abnormal sleep-study results. Children with sleep problems and a ferritin concentration $<50$ $\mu$g/L may benefit from iron supplementation.[70,71] Discuss obesity as a risk factor of sleep apnea and review need to implement healthy diet and activity in affected patients (see Health Supervision From 1 to 5 Years for discussion).

### Anticipatory Guidance From 5 to 12 Years at Every Health Supervision Visit, Unless Otherwise Indicated

- Review the child's development and appropriateness of transition to elementary school placement and any additional developmental intervention.
- Discuss socialization, family status, and relationships, including financial arrangements, health insurance, and guardianship, incorporating supported decision-making where recognized (see Resources for Families).
- Encourage development of age-appropriate social skills, self-help skills, and development of a sense of responsibility.
- Counsel families regarding the transition from elementary to middle school, when major change often occurs, from 1 teacher to several and from 1 class to changing classes. Prepare them to facilitate adjustment at a time when the academic disparity becomes greater and full inclusion becomes more difficult. Although transition to work-environment planning occurs formally at age 14 in the individualized education program, the discussion and participation in community resources may begin at a much earlier age, approximately age 10, and all subsequent visits.
- Refer patients with behavior or history concerning for autism for appropriate evaluation (see

Health Supervision From 1 to 5 Years for discussion).

- Continue to assess, monitor, and encourage independence with hygiene and self-care. Encourage family to teach, model, and respect privacy at home and in the community. Discuss appropriate management of sexual behaviors such as masturbation.
- Discuss progression of physical and psychosocial changes through puberty and issues of fertility and contraception.[118] Remind family that pubertal development usually follows patterns similar to those found in the general population, but the child with Down syndrome will likely need more preparation in understanding and managing these changes.[123]
- On at least 1 health supervision visit, educate family about increased risk of sexual exploitation (see "Anticipatory Guidance From 1 to 5 Years" for discussion).
- Discuss the need for gynecologic care in the pubescent girl. Provide developmentally appropriate discussion about puberty and include menses and dysmenorrhea (see Resources for Families). When developmentally appropriate on at least 1 visit, talk with the patient and her family about the chance of Down syndrome in her children (50%) if she were to become pregnant.[124]
- Although males with Down syndrome are usually infertile, there have been rare instances in which a male has reproduced.[125,126]
- When developmentally appropriate, birth control and prevention of sexually transmitted infections should be discussed with patients and their families.[123] Advocate for and offer long-acting, reversible contraception. Be familiar with local law and resources to assist the family in their decision-making regarding questions about long-term and reversible birth control.[123,127,128]
- At least once between 5 and 12 years of age, as with discussion in the first year of life, discuss future parental pregnancy planning and review risk possibility of recurrence of Down syndrome, as well as availability of prenatal testing options. Offer referral for genetic counseling if desired by the family.
- Parents should be advised that trampoline use should be avoided by all children, with or without Down syndrome, unless part of a structured training program with appropriate supervision and safety measures in place. Parents can be advised that participation in contact sports, such as football, soccer, and gymnastics, places children at risk for spinal cord injury.96,129
- Special Olympics requires documentation of physical examination of all athletes for participation in sports.[94]
- Be prepared to discuss and answer questions regarding treatments that are considered complementary or alternative (see Health Supervision From 1 Month to 1 Year for discussion).

## HEALTH SUPERVISION FROM 12 TO 21 YEARS OR OLDER: ADOLESCENCE TO EARLY ADULTHOOD

Follow *Bright Futures* schedule or more frequently as indicated.

### Physical Examination and Laboratory Values

- Obtain a history and perform a physical examination.
- Monitor weight and follow BMI trends at each health care visit. Review the adolescent/young adult's weight and height and plot it on the Down-syndrome specific charts for weight, height, and head circumference. These should be used in conjunction with the Centers for Disease Control BMI chart[58,120] (see "Health Supervision From 5 to 12 Years" for discussion). Counsel regarding healthy diet and a structured exercise program.
- Review feeding, ask if there have been any changes in eating patterns or respiratory symptoms with feeding, and ensure adequate iron intake (see Health Supervision From Birth to 1 Month for discussion).
- Emphasize healthy diet and lifestyle for preventing obesity.
- Obtain a CBC with differential and either (1) a combination of ferritin and CRP, or (2) a combination of serum iron and TIBC, beginning at 1 year of age and annually thereafter.
- A physician may elect to prescribe iron supplementation for adolescents/early adults with restless sleep and a ferritin concentration $<50$ μg/L for children or 75 for adults[71], (see Health Supervision From 1 to 5 Years for discussion).
- Palpate testes at each health supervision visit (see Health Supervision From 1 to 5 Years for discussion).
- Measure TSH concentration annually and obtain TSH sooner if there are symptoms of thyroid dysfunction (see Health Supervision From Birth to 1 Month for discussion). Measure TSH every 6 months if antithyroid antibodies have been detected.
- Obtain annual ear-specific audiologic evaluation (see Health Supervision From 1 Month to 1 Year for discussion). If middle ear disease occurs, obtain developmentally appropriate hearing evaluation after treatment.
- For adolescents/early adults on a diet that contains gluten, review for symptoms potentially related to celiac disease at every health

supervision visit and evaluate if indicated (see Health Supervision From 1 to 5 Years for discussion).
- Individualize cardiology follow-up on the basis of history of cardiac defects.
- Discuss symptoms related to sleep-disordered breathing, including snoring, restless sleep, daytime sleepiness, nighttime awakening, behavior problems, and sleep position, at every health supervision visit. Refer to a physician with expertise in pediatric sleep any child with signs or symptoms of sleep-disordered breathing or an abnormal sleep-study result. Discuss the risk factor of obesity for sleep apnea and counsel regarding healthy diet and activity if needed.
- Discuss with caregivers and the patient at every health supervision visit the importance of cervical spine-positioning precautions for protection of the cervical spine during any anesthetic, surgical, or radiographic procedure. Perform careful history and physical examination with attention to myelopathic signs and symptoms. Caregivers and patients should also be instructed to contact their physician immediately for new onset of symptoms of myelopathy (see Health Supervision From 1 to 5 Years for discussion).
- Inquire regarding symptoms of neurologic dysfunction, including seizures, and perform neurologic examination.
- Discuss behavioral and social status and refer patients who have depression,[130] chronic behavioral problems, or acute deterioration in function for specialized evaluation and intervention.[112,113,121]
- Inquire regarding symptoms of acute regression (see Health Supervision From 1 to 5 Years for discussion).
- Skin problems are particularly common in people with Down syndrome. Discuss skin, hair, and scalp care at each preventive health care visit and consider referral to a dermatologist if needed (see Health Supervision From 1 to 5 Years for discussion). In addition, inflammatory disorders such as hidradenitis suppurativa may present at an older age.[131]
- Perform visual acuity testing or photoscreening, if available, at every health supervision visit or ensure adolescent/young adult is under care of a pediatric ophthalmologist or ophthalmologist experienced in care of people with disabilities who will determine the frequency of assessment. Assessment for onset of cataracts, refractive errors, and keratoconus, which can cause blurred vision, corneal thinning, or corneal haze and is typically diagnosed after puberty, is important.[63,85]
- Examine annually for acquired mitral and aortic valvular disease in older patients with Down syndrome. An echocardiogram should be obtained if there is a history of increasing fatigue, shortness of breath, exertional dyspnea, or a new murmur or gallop.
- Refer patients with behavior or history concerning for autism spectrum disorder for appropriate evaluation and therapy (see Anticipatory Guidance From 1 to 5 Years for discussion).

### Anticipatory Guidance From 12 to 21 Years and Older at Every Health Supervision Visit, Unless Otherwise Indicated

- Issues related to transition into adulthood, including educational goals, work, independence, and transition of medical care. Continue these discussions and include guardianship incorporating supported decision-making, where recognized, and long-term financial planning from early adolescence (see Resources for Families).[132,133] Potential adult morbidities, including apparent tendency toward premature aging and increased risk of Alzheimer disease, may also be discussed.[134]
- Discuss appropriateness of school placement and vocational planning as early as possible in the school setting and emphasize planning for transition to adulthood and adequate vocational training within the school curriculum.
- Patients/caregivers should be advised that trampoline use should be avoided by all children, with or without Down syndrome, unless part of a structured training program with appropriate supervision and safety measures in place. Parents can be advised that participation in contact sports, such as football, soccer, and gymnastics, places children at risk for spinal cord injury.[129]
- Special Olympics requires documentation of physical examination of all athletes for participation in sports.[94] Be prepared to discuss and answer questions regarding treatments that are considered complementary or alternative (see Health Supervision From 1 Month to 1 Year for discussion).
- On at least 1 health supervision visit, educate family about increased risk of sexual exploitation (see Anticipatory Guidance From 1 to 5 Years for discussion).
- Continue to assess, monitor, and encourage independence with hygiene and self-care. Provide guidance on healthy, normal, and typical sexual development and behaviors. Provide education and guidance about normal masturbation behaviors and personal boundaries. Emphasize the need

for understandable information and encourage opportunities for advancing comprehension of sexuality (see Resources for Families). Discuss the need for contraception and prevention of sexually transmitted infections and the degree of supervision required. Advocate for and offer use of long-acting, reversible contraception and be familiar with local laws and resources to assist the family in their decision-making regarding questions about long-term and reversible birth control.[123]

- Make recommendations and provide or refer for routine gynecologic care as needed for long-acting, reversible contraception or other indications if not already provided. Discuss premenstrual behavioral problems and management of menses, including caregiver concerns regarding menstrual suppression for hygiene purposes.[128,135,136]
- At least at 1 visit, talk with the female patient and her family about the chance that she could have a child with Down syndrome if she were to become pregnant.[123–124]
- Discuss independent living opportunities, group homes, options for postsecondary education, workshop settings, and other community-supported employment.
- Discuss intrafamily relationships, financial planning including the Achieving a Better Life Experience Act (see Resources for Families), and guardianship including supported decision-making where recognized.[132]
- Facilitate transition and provide coordination to adult medical primary and subspecialty care.[137] It is recognized that many young adults receive care from pediatricians, and providers will want to be aware of the newly developed health supervision for adults.[138]

## FUTURE CONSIDERATIONS

Many issues related to the development and health of people with Down syndrome remain to be evaluated, and research agendas for addressing both public health and basic science topics have been developed. Knowledge in several topics of great importance to the care of children with Down syndrome could be enhanced through population-based research. A rigorous, evidence-based review of screening and treatment of atlantoaxial instability, for example, is needed, and continuing research is critical for directing the care for optimal outcomes of people with Down syndrome.[1,139–141]

## RESOURCES FOR FAMILIES

### Prenatal and Infancy

- Brighter Tomorrows Supporting Families: https://hdi.uky.edu/project/brighter-tomorrows-supporting-families-with-accurate-information-about-down-syndrome. Supporting families with accurate information about Down syndrome (includes Lettercase resources). Interdisciplinary Human Development Institute, University of Kentucky.
- Lettercase resources: www.lettercase.org. Provides prenatal and postnatal counseling for families. One copy provided free to professionals and family.
- Down Syndrome Diagnosis Network: www.dsdiagnosisnetwork.org. Cohorts of families with similar due dates or birthdates connected in moderated Facebook groups.
- Skallerup SJ. *Babies With Down Syndrome: A New Parents Guide*, 3rd ed. Bethesda, MD: Woodbine House; 2009. (English and Spanish editions available at www.woodbinehouse.com.) Provides guidance for raising and caring for a child with Down syndrome through age 5.

### Childhood

- Pueschel SM, ed. *A Parent's Guide to Down Syndrome: Toward a Brighter Future*. Bethesda, MD: Brookes Publishing; 2001.
- Stein DS. *Supporting Positive Behavior in Children and Teens with Down Syndrome: The Respond but Don't React Method*. Bethesda, MD: Woodbine House; 2016.
- www.woodbinehouse.com. Variety of books for families, therapists, and teachers of children with Down syndrome.

### Adolescence

- Got Transition: http://www.gottransition.org/. Provides resources and guidance for transition.
- Couwenhoven T. *Boys Guide to Growing Up–Choices & Changes During Puberty Written for Persons with Intellectual Disability*. Bethesda, MD: Woodbine House; 2012.
- Couwenhoven T. *Girls Guide to Growing Up–Choices & Changes During Puberty Written for Persons with Intellectual Disability*. Bethesda, MD: Woodbine House; 2012.
- Simmons J. *The Down Syndrome Transition Handbook: Charting Your Child's Course to Adulthood*. Bethesda, MD: Woodbine House; 2010. Available at: www.woodbinehouse.com.

### Across the Lifespan

- DS-Connect: The Down Syndrome Registry: https://DSConnect.nih.gov. Families and patients connect with researchers and health care providers and may participate in clinical studies and take confidential health-related surveys that help achieve better understanding of people with Down syndrome across the lifespan.
- National Down Syndrome Congress: www.ndsccenter.org. Information, advocacy, education, and

support for persons with Down syndrome, siblings, and families in English and Spanish.
- National Down Syndrome Society: www.ndss.org. Advocacy and information regarding Down syndrome across the lifespan.
- Family Voices: https://familyvoices.org/affiliates/. Family-to-family health information centers that help families navigate systems needed by children with special health care needs.
- Parent to Parent USA: https://www.p2pusa.org/parents. Provides support to deal with challenges of raising children with special health care needs.
- Parent Training and Information Centers: https://www.parentcenterhub.org. Resources in each state that inform, prepare, and assist families with navigation of the education system.
- March of Dimes: www.marchofdimes.com. Information for parents on health issues related to pregnancy and birth defects.
- Down Syndrome Education International: www.downsed.org. Resources for educators and parents relevant to communication, numeracy, speech, and supporting inclusion.
- Canadian Down Syndrome Society: www.cdss.ca. Resources for families on a variety of issues related to Down syndrome.
- ABLE ACT 2014: https://www.irs.gov/government-entities/federal-state-local-governments/able-accounts-tax-benefit-for-people-with-disabilities. Tax benefit for people with disabilities: allow states to create tax-advantage savings programs for eligible people with disabilities (designated beneficiaries) and can help pay for qualified disability expenses.
- DSC2U Down syndrome clinic to you: www.dsc2u.org. Provides personalized health and wellness information about Down syndrome to caregivers and primary care physicians.


### Lead Authors

Marilyn J. Bull, MD, FAAP
Tracy Trotter, MD, FAAP
Stephanie L. Santoro, MD, FAAP
Celanie Christensen, MD, MS, FAAP
Randall W. Grout, MD, MS, FAAP

### Council on Genetics Executive Committee, 2019–2020

Leah W. Burke, MD, FAAP; Chairperson
Susan A. Berry, MD, FAAP
Timothy A. Geleske, MD, FAAP
Ingrid Holm, MD, FAAP
Robert J. Hopkin, MD, FAAP
Wendy J. Introne, MD, FAAP
Michael J. Lyons, MD, FAAP
Danielle C. Monteil, MD, FAAP
Angela Scheuerle, MD, FAAP
Joan M. Stoler, MD, FAAP
Samantha A. Vergano, MD, FAAP

### Former Executive Committee Members

Emily Chen, MD, PhD, FAAP, Co-Chairperson
Rizwan Hamid, MD, PhD, FAAP
Tracy L. Trotter, MD, FAAP, Co-Chairperson

### Partnership for Policy Implementation Contributors

Stephen M. Downs, MD, MS, FAAP
Randall W. Grout, MD, MS, FAAP

### Liaisons

Christopher Cunniff, MD, PhD, FAAP; American College of Medical Genetics
Melissa A. Parisi, MD, PhD, FAAP; Eunice Kennedy Shriver National Institute of Child Health and Human Development
Steven J. Ralston, MD; American College of Obstetricians and Gynecologists
Joan A. Scott, MS, CGC; Health Resources and Services Administration, Maternal and Child Health Bureau
Stuart K. Shapira, MD, PhD; Centers for Disease Control and Prevention

### Staff

Paul Spire


### ABBREVIATIONS

BAER: brainstem auditory evoked response
CBC: complete blood cell count
cfDNA: cell-free DNA
CRP: C-reactive protein
CVS: chorionic villus sampling
FISH: fluorescent in situ hybridization
IgA: immunoglobulin A
T4: free thyroxine
TAM: transient abnormal myelopoiesis
TIBC: total iron-binding capacity
TSH: thyroid-stimulating hormone
TTG: tissue transglutaminase

### REFERENCES


1. Schieve LA, Boulet SL, Boyle C, Rasmussen SA, Schendel D. Health of children 3 to 17 years of age with Down syndrome in the 1997-2005 national health interview survey. *Pediatrics*. 2009;123(2):e253–e260
2. Roizen NJ, Magyar CI, Kuschner ES, et al. A community cross-sectional survey of medical problems in 440 children with Down syndrome in New York State. *J Pediatr*. 2014;164(4):871–875
3. Stoll C, Dott B, Alembik Y, Roth MP. Associated congenital anomalies among cases with Down syndrome. *Eur J Med Genet*. 2015;58(12):674–680
4. Shaw EDBR, Beals RK. The hip joint in Down syndrome. A study of its structure and associated disease. *Clin Orthop Relat Res*. 1992; (278): 101–107

5. Caird MS, Wills BP, Dormans JP. Down syndrome in children: the role of the orthopaedic surgeon. *J Am Acad Orthop Surg*. 2006;14(11):610–619

6. Pueschel SM, Findley TW, Furia J, Gallagher PL, Scola FH, Pezzullo JC. Atlantoaxial instability in Down syndrome: roentgenographic, neurologic, and somatosensory evoked potential studies. *J Pediatr*. 1987;110(4):515–521

7. Selby KA, Newton RW, Gupta S, Hunt L. Clinical predictors and radiological reliability in atlantoaxial subluxation in Down syndrome. *Arch Dis Child*. 1991;66(7):876–878

8. Pierce MJ, LaFranchi SH, Pinter JD. Characterization of thyroid abnormalities in a large cohort of children with Down syndrome. *Horm Res Paediatr*. 2017;87(3):170–178

9. Juj H, Emery H. The arthropathy of Down syndrome: an underdiagnosed and under-recognized condition. *J Pediatr*. 2009;154(2):234–238

10. Bergholdt R, Eising S, Nerup J, Pociot F. Increased prevalence of Down syndrome in individuals with type 1 diabetes in Denmark: a nationwide population-based study. *Diabetologia*. 2006;49(6):1179–1182

11. Kent L, Evans J, Paul M, Sharp M. Comorbidity of autistic spectrum disorders in children with Down syndrome. *Dev Med Child Neurol*. 1999;41(3):153–158

12. Moss J, Richards C, Nelson L, Oliver C. Prevalence of autism spectrum disorder symptomatology and related behavioural characteristics in individuals with Down syndrome. *Autism*. 2013;17(4):390–404

13. Hook EB. *Epidemiology of Down Syndrome*. Cambridge: Ware Press; 1982

14. Bull MJ. Down syndrome. *N Engl J Med*. 2020;382(24):2344–2352

15. Papavassiliou P, Charalsawadi C, Rafferty K, Jackson-Cook C. Mosaicism for trisomy 21: a review. *Am J Med Genet A*. 2015;167A(1):26–39

16. Pelleri MC, Cicchini E, Locatelli C, et al. Systematic reanalysis of partial trisomy 21 cases with or without Down syndrome suggests a small region on 21q22.13 as critical to the phenotype. *Hum Mol Genet*. 2016;25(12):2525–2538

17. ACOG Committee on Practice Bulletins. ACOG practice bulletin no. 77: screening for fetal chromosomal abnormalities. *Obstet Gynecol*. 2007;109(1):217–227

18. American College of Obstetricians and Gynecologists. ACOG practice bulletin no. 88, December 2007. Invasive prenatal testing for aneuploidy. *Obstet Gynecol*. 2007;110(6):1459–1467

19. Gil MM, Accurti V, Santacruz B, Plana MN, Nicolaides KH. Analysis of cell-free DNA in maternal blood in screening for aneuploidies: updated meta-analysis. *Ultrasound Obstet Gynecol*. 2017;50(3):302–314

20. American College of Obstetricians and Gynecologists' Committee on Practice Bulletins–Obstetrics; Committee on Genetics; Society for Maternal-Fetal Medicine. Screening for fetal chromosomal abnormalities: ACOG Practice Bulletin, Number 226. *Obstet Gynecol*. 2020;136(4):e48–e69

21. Driscoll DA, Gross SJ. Screening for fetal aneuploidy and neural tube defects. *Genet Med*. 2009;11(11):818–821

22. Malone FD, Canick JA, Ball RH, et al. First- and Second-Trimester Evaluation of Risk (FASTER) Research Consortium. First-trimester or second-trimester screening, or both, for Down syndrome. *N Engl J Med*. 2005;353(19):2001–2011

23. Wald NJ, Rodeck C, Hackshaw AK, Walters J, Chitty L, Mackinson AM; SURUSS Research Group. First and second trimester antenatal screening for Down syndrome: the results of the Serum, Urine and Ultrasound Screening Study (SURUSS). *Health Technol Assess*. 2003;7(11):1–77

24. Spencer K, Spencer CE, Power M, Dawson C, Nicolaides KH. Screening for chromosomal abnormalities in the first trimester using ultrasound and maternal serum biochemistry in a one-stop clinic: a review of three years prospective experience. *BJOG*. 2003;110(3):281–286

25. [No authors listed] Practice bulletin no. 162: prenatal diagnostic testing for genetic disorders. *Obstet Gynecol*. 2016;127(5):e108–e122

26. Akolekar R, Beta J, Picciarelli G, Ogilvie C, D'Antonio F. Procedure-related risk of miscarriage following amniocentesis and chorionic villus sampling: a systematic review and meta-analysis. *Ultrasound Obstet Gynecol*. 2015;45(1):16–26

27. Adams RC, Levy SE. Council on Children With Disabilities. Shared decision-making and children with disabilities: pathways to consensus. *Pediatrics*. 2017;139(6):e20170956

28. Hornberger LL. Committee on Adolescence. Options counseling for the pregnant adolescent patient. *Pediatrics*. 2017;140(3):e20172274

29. Guseh SH, Little SE, Bennett K, Silva V, Wilkins-Haug LE. Antepartum management and obstetric outcomes among pregnancies with Down syndrome from diagnosis to delivery. *Prenat Diagn*. 2017;37(7):640–646

30. Skotko BG, Capone GT, Kishnani PS. Down Syndrome Diagnosis Study Group. Postnatal diagnosis of Down syndrome: synthesis of the evidence on how best to deliver the news. *Pediatrics*. 2009;124(4):e751–e758

31. Skotko BG, Levine SP, Goldstein R. Self-perceptions from people with Down syndrome. *Am J Med Genet A*. 2011;155A(10):2360–2369

32. Crocker AF, Smith SN. Person-first language: are we practicing what we preach? *J Multidiscip Healthc*. 2019;12:125–129

33. Skotko BG, Levine SP, Goldstein R. Having a brother or sister with Down syndrome: perspectives from siblings. *Am J Med Genet A*. 2011;155A(10):2348–2359

34. Skotko BG, Levine SP, Macklin EA, Goldstein RD. Family perspectives about Down syndrome. *Am J Med Genet A*. 2016;170A(4):930–941

35. Jackson A, Maybee J, Moran MK, Wolter-Warmerdam K, Hickey F. Clinical characteristics of dysphagia in children with Down syndrome. *Dysphagia*. 2016;31(5):663–671

36. Romano C, van Wynckel M, Hulst J, et al. European Society for Paediatric Gastroenterology, hepatology and nutrition guidelines for the evaluation and treatment of gastrointestinal and

nutritional complications in children with neurological impairment. *J Pediatr Gastroenterol Nutr*. 2017;65(2):242–264

37. Stanley MA, Shepherd N, Duvall N, et al. Clinical identification of feeding and swallowing disorders in 0–6-month-old infants with Down syndrome. *Am J Med Genet A*. 2019;179(2):177–182
38. Poskanzer SA, Hobensack VL, Ciciora SL, Santoro SL. Feeding difficulty and gastrostomy tube placement in infants with Down syndrome. *Eur J Pediatr*. 2020;179(6):909–917
39. Jackson A, Maybee J, Wolter-Warmerdam K, DeBoer E, Hickey F. Associations between age, respiratory comorbidities, and dysphagia in infants with Down syndrome. *Pediatr Pulmonol*. 2019;54(11):1853–1859
40. McDowell KM, Craven DI. Pulmonary complications of Down syndrome during childhood. *J Pediatr*. 2011;158(2):319–325
41. Santoro SL, Atoum D, Hufnagel RB, Motley WW. Surgical, medical and developmental outcomes in patients with Down syndrome and cataracts. *SAGE Open Med*. 2017;5:2050312117715583
42. Haargaard B, Fledelius HC. Down syndrome and early cataract. *Br J Ophthalmol*. 2006;90(8):1024–1027
43. Muse C, Harrison J, Yoshinaga-Itano C, et al. Joint Committee on Infant Hearing of the American Academy of Pediatrics. Supplement to the JCIH 2007 position statement: principles and guidelines for early intervention after confirmation that a child is deaf or hard of hearing. *Pediatrics*. 2013;131(4):e1324–e1349
44. Park AH, Wilson MA, Stevens PT, Harward R, Hohler N. Identification of hearing loss in pediatric patients with Down syndrome. *Otolaryngol Head Neck Surg*. 2012;146(1):135–140
45. Bull MJ, Engle WA. Committee on Injury, Violence, and Poison Prevention and Committee on Fetus and Newborn; American Academy of Pediatrics. Safe transportation of preterm and low birth weight infants at hospital discharge. *Pediatrics*. 2009;123(5):1424–1429
46. Roberts I, Alford K, Hall G, et al. Oxford-Imperial Down Syndrome Cohort Study Group. GATA1-mutant clones are frequent and often unsuspected in babies with Down syndrome: identification of a population at risk of leukemia. *Blood*. 2013;122(24):3908–3917
47. Tunstall O, Bhatnagar N, James B, et al. British Society for Haematology. Guidelines for the investigation and management of transient leukaemia of Down Syndrome. *Br J Haematol*. 2018;182(2):200–211
48. Kivivuori SM, Rajantie J, Siimes MA. Peripheral blood cell counts in infants with Down syndrome. *Clin Genet*. 1996;49(1):15–19
49. Hasle H, Clemmensen IH, Mikkelsen M. Risks of leukaemia and solid tumours in individuals with Down syndrome. *Lancet*. 2000;355(9199):165–169
50. Iughetti L, Predieri B, Bruzzi P, et al. Ten-year longitudinal study of thyroid function in children with Down syndrome. *Horm Res Paediatr*. 2014;82(2):113–121
51. Lagan N, Huggard D, Mc Grane F, et al. Multiorgan involvement and management in children with Down syndrome. *Acta Paediatr*. 2020;109(6):1096–1111
52. Santoro SL, Chicoine B, Jasien JM, et al. Pneumonia and respiratory infections in Down syndrome: a scoping review of the literature. *Am J Med Genet A*. 2021;185(1):286–299
53. American Academy of Pediatrics. Respiratory syncytial virus. In: Kimberlin DW, Barnett ED, Lynfield R, Sawyer MH, eds. *Red Book: Report of the Committee on Infectious Diseases* 2021. 32nd ed. Itasca, IL: American Academy of Pediatrics; 2021:628–636
54. Lewanda AF, Gallegos MF, Summar M. Patterns of dietary supplement use in children with Down syndrome. *J Pediatr*. 2018;201:100–105.e30
55. McClafferty H, Vohra S, Bailey M, et al. Section on Integrative Medicine. Pediatric Integrative Medicine. *Pediatrics*. 2017;140(3):e20171961
56. Kupferman JC, Druschel CM, Kupchik GS. Increased prevalence of renal and urinary tract anomalies in children with Down syndrome. *Pediatrics*. 2009;124(4):e615–e621
57. Zemel BS, Pipan M, Stallings VA, et al. Growth charts for children with Down syndrome in the United States. *Pediatrics*. 2015;136(5):e1204–e1211
58. Hatch-Stein JA, Zemel BS, Prasad D, et al. Body composition and BMI growth charts in children with Down syndrome. *Pediatrics*. 2016;138(4):e20160541
59. Shott SR. Down syndrome: common otolaryngologic manifestations. *Am J Med Genet C Semin Med Genet*. 2006;142C(3):131–140
60. De Schrijver L, Topsakal V, Wojciechowski M, Van de Heyning P, Boudewyns A. Prevalence and etiology of sensorineural hearing loss in children with down syndrome: a cross-sectional study. *Int J Pediatr Otorhinolaryngol*. 2019;116:168–172
61. Blaser S, Propst EJ, Martin D, et al. Inner ear dysplasia is common in children with Down syndrome (trisomy 21). *Laryngoscope*. 2006;116(12):2113–2119
62. American Academy of Ophthalmology. Trisomy 21/Down syndrome. Available at: https://eyewiki.aao.org/TGrisomy_21/Down_Syndrome. Accessed August 7, 2020
63. Umfress AC, Hair CD, Donahue SP. Prevalence of ocular pathology on initial screening and incidence of new findings on follow-up examinations in children with trisomy 21. *Am J Ophthalmol*. 2019;207:373–377
64. AAPOS. Vision screening recommendation. Available at: https://aapos.org/patients/patient-resources. 2019. Accessed August 7, 2020
65. Coats DK, McCreery KM, Plager DA, Bohra L, Kim DS, Paysse EA. Nasolacrimal outflow drainage anomalies in Down's syndrome. *Ophthalmology*. 2003;110(7):1437–1441
66. Martin T, Smith A, Breatnach CR, et al. Infants born with Down syndrome: burden of disease in the early neonatal period. *J Pediatr*. 2018;193:21–26
67. Dixon NE, Crissman BG, Smith PB, Zimmerman SA, Worley G, Kishnani PS. Prevalence of iron deficiency in children with Down syndrome. *J Pediatr*. 2010;157(6):967–971.e1

68. Georgieff MK. Long-term brain and behavioral consequences of early iron deficiency. *Nutr Rev*. 2011;69(Suppl 1):S43–S48

69. Hart SJ, Zimmerman K, Linardic CM, et al. Detection of iron deficiency in children with Down syndrome. *Genet Med*. 2020;22(2):317–325

70. Dosman C, Witmans M, Zwaigenbaum L. Iron's role in paediatric restless legs syndrome – a review. *Paediatr Child Health*. 2012;17(4):193–197

71. Allen RP, Picchietti DL, Auerbach M, et al. International Restless Legs Syndrome Study Group (IRLSSG). Evidence-based and consensus clinical practice guidelines for the iron treatment of restless legs syndrome/Willis-Ekbom disease in adults and children: an IRLSSG task force report. *Sleep Med*. 2018;41:27–44

72. Taub JW, Berman JN, Hitzler JK, et al. Improved outcomes for myeloid leukemia of Down syndrome: a report from the Children's Oncology Group AAML0431 trial. *Blood*. 2017;129(25):3304–3313

73. Matloub Y, Rabin KR, Ji L, et al. Excellent long-term survival of children with Down syndrome and standard-risk ALL: a report from the Children's Oncology Group. *Blood Adv*. 2019;3(11):1647–1656

74. Goldberg-Stern H, Strawsburg RH, Patterson B, et al. Seizure frequency and characteristics in children with Down syndrome. *Brain Dev*. 2001;23(6):375–378

75. Kumada T, Ito M, Miyajima T, et al. Multi-institutional study on the correlation between chromosomal abnormalities and epilepsy. *Brain Dev*. 2005;27(2):127–134

76. Jea A, Smith ER, Robertson R, Scott RM. Moyamoya syndrome associated with Down syndrome: outcome after surgical revascularization. *Pediatrics*. 2005;116(5):e694–e701

77. See AP, Ropper AE, Underberg DL, Robertson RL, Scott RM, Smith ER. Down syndrome and moyamoya: clinical presentation and surgical management. *J Neurosurg Pediatr*. 2015;16(1):58–63

78. Centers for Disease Control and Prevention. Immunization schedules. Available at: https://www.cdc.gov/vaccines/schedules. Accessed December 14, 2020

79. Fitzgerald DA, Paul A, Richmond C. Severity of obstructive apnoea in children with Down syndrome who snore. *Arch Dis Child*. 2007;92(5):423–425

80. Shott SR, Amin R, Chini B, Heubi C, Hotze S, Akers R. Obstructive sleep apnea: should all children with Down syndrome be tested? *Arch Otolaryngol Head Neck Surg*. 2006;132(4):432–436

81. Kobayashi T, Sakemi Y, Yamashita H. Increased incidence of retroperitoneal teratomas and decreased incidence of sacrococcygeal teratomas in infants with Down syndrome. *Pediatr Blood Cancer*. 2014;61(2):363–365

82. Satgé D, Vekemans M. Down syndrome patients are less likely to develop some (but not all) malignant solid tumours. *Clin Genet*. 2011;79(3):289–290, author reply 291–292

83. Hasle H, Friedman JM, Olsen JH, Rasmussen SA. Low risk of solid tumors in persons with Down syndrome. *Genet Med*. 2016;18(11):1151–1157

84. Lin K, Sharangpani R. Screening for testicular cancer: an evidence review for the U.S. Preventive Services Task Force. *Ann Intern Med*. 2010;153(6):396–399

85. Yanovitch T, Wallace DK, Freedman SF, et al. The accuracy of photoscreening at detecting treatable ocular conditions in children with Down syndrome. *J AAPOS*. 2010;14(6):472–477

86. Berk AT, Saatci AO, Erçal MD, Tunç M, Ergin M. Ocular findings in 55 patients with Down's syndrome. *Ophthalmic Genet*. 1996;17(1):15–19

87. Locke GR, Gardner JI, Van Epps EF. Atlas-dens interval (ADI) in children: a survey based on 200 normal cervical spines. *Am J Roentgenol Radium Ther Nucl Med*. 1966;97(1):135–140

88. Burke SW, French HG, Roberts JM, Johnston CE II, Whitecloud TS III, Edmunds JO Jr. Chronic atlanto-axial instability in Down syndrome. *J Bone Joint Surg Am*. 1985;67(9):1356–1360

89. Morton RE, Khan MA, Murray-Leslie C, Elliott S. Atlantoaxial instability in Down's syndrome: a five year follow up study. *Arch Dis Child*. 1995;72(2):115–118, discussion 118–119

90. Hengartner AC, Whelan R, Maj R, Wolter-Warmerdam K, Hickey F, Hankinson TC. Evaluation of 2011 AAP cervical spine screening guidelines for children with Down Syndrome. *Childs Nerv Syst*. 2020;36(11):2609–2614

91. Pueschel SM, Scola FH, Pezzullo JC. A longitudinal study of atlanto-dens relationships in asymptomatic individuals with Down syndrome. *Pediatrics*. 1992;89(6 Pt 2):1194–1198

92. Ferguson RL, Putney ME, Allen BL Jr. Comparison of neurologic deficits with atlanto-dens intervals in patients with Down syndrome. *J Spinal Disord*. 1997;10(3):246–252

93. Nader-Sepahi A, Casey AT, Hayward R, Crockard HA, Thompson D. Symptomatic atlantoaxial instability in Down syndrome. *J Neurosurg*. 2005;103(3 Suppl):231–237

94. Special Olympics. Athlete Medical Form – Health History. Available at: http://media.specialolympics.org/resources/health/disciplines/medfest/MedFest-Health-History-and-Physical-Exam-Form-NON-US-Programs-fillable.pdf. Accessed March 25, 2022

95. Brockmeyer D. Down syndrome and craniovertebral instability. Topic review and treatment recommendations. *Pediatr Neurosurg*. 1999;31(2):71–77

96. Briskin S, LaBotz M. Council on Sports Medicine and Fitness; American Academy of Pediatrics. Trampoline safety in childhood and adolescence. *Pediatrics*. 2012;130(4):774–779. Reaffirmed July 2015, March 2020

97. Hill ID, Dirks MH, Liptak GS, et al. North American Society for Pediatric Gastroenterology, Hepatology and Nutrition. Guideline for the diagnosis and treatment of celiac disease in children: recommendations of the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition. *J Pediatr Gastroenterol Nutr*. 2005;40(1):1–19

98. Bonamico M, Mariani P, Danesi HM, et al. SIGEP (Italian Society of Pediatric Gastroenterology and Hepatology) and Medical Genetic Group. Prevalence and clinical picture of celiac disease in italian down syndrome patients: a multicenter study. *J Pediatr Gastroenterol Nutr*. 2001;33(2):139–143

99. Swigonski NL, Kuhlenschmidt HL, Bull MJ, Corkins MR, Downs SM. Screening for celiac disease in asymptomatic

children with Down syndrome: cost-effectiveness of preventing lymphoma. *Pediatrics*. 2006;118(2):594–602

100. Ng DK, Chan CH, Cheung JM. Children with Down syndrome and OSA do not necessarily snore. *Arch Dis Child*. 2007;92(11):1047–1048
101. Bassett EC, Musso MF. Otolaryngologic management of Down syndrome patients: what is new? *Curr Opin Otolaryngol Head Neck Surg*. 2017;25(6):493–497
102. Farhood Z, Isley JW, Ong AA, et al. Adenotonsillectomy outcomes in patients with Down syndrome and obstructive sleep apnea. *Laryngoscope*. 2017;127(6):1465–1470
103. Bertapelli F, Pitetti K, Agiovlasitis S, Guerra-Junior G. Overweight and obesity in children and adolescents with Down syndrome-prevalence, determinants, consequences, and interventions: A literature review. *Res Dev Disabil*. 2016;57:181–192
104. Wolraich ML, Hagan JF Jr, Allan C, et al. Subcommittee on Children and Adolescents With Attention-Deficit/Hyperactive Disorder. Clinical practice guideline for the diagnosis, evaluation, and treatment of attention-deficit/hyperactivity disorder in children and adolescents. *Pediatrics*. 2019;144(4):e20192528
105. Molloy CA, Murray DS, Kinsman A, et al. Differences in the clinical presentation of trisomy 21 with and without autism. *J Intellect Disabil Res*. 2009;53(2):143–151
106. Rasmussen P, Börjesson O, Wentz E, Gillberg C. Autistic disorders in Down syndrome: background factors and clinical correlates. *Dev Med Child Neurol*. 2001;43(11):750–754
107. Kielinen M, Rantala H, Timonen E, Linna SL, Moilanen I. Associated medical disorders and disabilities in children with autistic disorder: a population-based study. *Autism*. 2004;8(1):49–60
108. Capone GT, Grados MA, Kaufmann WE, Bernad-Ripoll S, Jewell A. Down syndrome and comorbid autism-spectrum disorder: characterization using the aberrant behavior checklist. *Am J Med Genet A*. 2005;134(4):373–380
109. Carter JC, Capone GT, Gray RM, Cox CS, Kaufmann WE. Autistic-spectrum disorders in Down syndrome: further delineation and distinction from other behavioral abnormalities. *Am J Med Genet B Neuropsychiatr Genet*. 2007;144B(1):87–94
110. Dressler A, Perelli V, Bozza M, Bargagna S. The autistic phenotype in Down syndrome: differences in adaptive behaviour versus Down syndrome alone and autistic disorder alone. *Funct Neurol*. 2011;26(3):151–158
111. Hyman SL, Levy SE, Myers SM. Council on Children With Disabilities, Section on Developmental and Behavioral Pediatrics. Identification, evaluation, and management of children with autism spectrum disorder. *Pediatrics*. 2020;145(1):e20193447
112. Prasher V. Disintegrative syndrome in young adults. *Ir J Psychol Med*. 2002;19(3):101
113. Worley G, Crissman BG, Cadogan E, Milleson C, Adkins DW, Kishnani PS. Down syndrome disintegrative disorder: new-onset autistic regression, dementia, and insomnia in older children and adolescents with Down syndrome. *J Child Neurol*. 2015;30(9):1147–1152
114. Pikora TJ, Bourke J, Bathgate K, Foley KR, Lennox N, Leonard H. Health conditions and their impact among adolescents and young adults with Down syndrome. *PLoS One*. 2014;9(5):e96868
115. Rork JF, McCormack L, Lal K, Wiss K, Belazarian L. Dermatologic conditions in Down syndrome: a single-center retrospective chart review. *Pediatr Dermatol*. 2020;37(5):811–816
116. Chow KM, O'Donnell D. Concomitant occurrence of hypodontia and supernumerary teeth in a patient with Down syndrome. *Spec Care Dentist*. 1997;17(2):54–57
117. Andersson EM, Axelsson S, Austeng ME, et al. Bilateral hypodontia is more common than unilateral hypodontia in children with Down syndrome: a prospective population-based study. *Eur J Orthod*. 2014;36(4):414–418
118. Couwenhoven T. *Teaching Children with Down Syndrome about Their Bodies, Boundaries, and Sexuality (Topics in Down Syndrome)*. Bethesda, MD: Woodbine House, Inc.; 2007
119. Stein, DS. *Supporting Positive Behavior in Children and Teens With Syndrome: The Respond but Don't React Method*. Bethesda, MD: Woodbine House; 2016.
120. Centers for Disease Control and Prevention (CDC). Growth Charts for Children with Down Syndrome. Available at: https://www.cdc.gov/ncbddd/birthdefects/downsyndrome/growthcharts.html. Accessed March 25, 2022
121. Brodtmann A. Hashimoto encephalopathy and Down syndrome. *Arch Neurol*. 2009;66(5):663–666
122. Palumbo ML, McDougle CJ. Pharmacotherapy of Down syndrome. *Expert Opin Pharmacother*. 2018;19(17):1875–1889
123. Centers for Disease Control and Prevention. CDC Contraceptive Guidance for Health Care Providers. Available at: https://www.cdc.gov/reproductivehealth/contraception/contraception_guidance.htm. Accessed March 25, 2022
124. Gardner RJMSG. *Chromosome Abnormalities and Genetic Counseling*, 3rd ed. New York, NY: Oxford University Press; 2004
125. Sheridan R, Llerena J Jr, Matkins S, Debenham P, Cawood A, Bobrow M. Fertility in a male with trisomy 21. *J Med Genet*. 1989;26(5):294–298
126. Pradhan M, Dalal A, Khan F, Agrawal S. Fertility in men with Down syndrome: a case report. *Fertil Steril*. 2006;86(6):1765.e1–1765.e3
127. Katz AL, Webb SA. Committee on Bioethics. Informed consent in decision-making in pediatric practice. *Pediatrics*. 2016;138(2):e20161485
128. Burke LM, Kalpakjian CZ, Smith YR, Quint EH. Gynecologic issues of adolescents with Down syndrome, autism, and cerebral palsy. *J Pediatr Adolesc Gynecol*. 2010;23(1):11–15
129. Carbone PSSP, Smith PJ, Lewis C, LeBlanc C. Promoting the participation of children and adolescents with disabilities in sports, recreation, and physical activity. *Pediatrics*. 2021;148(6):e2021054664
130. Zuckerbrot RA, Cheung A, Jensen PS, Stein REK, Laraque D. GLAD-PC Steering

Group. Guidelines for adolescent depression in primary care (GLAD-PC): part I. Practice preparation, identification, assessment, and initial management. *Pediatrics*. 2018;141(3): e20174081

131. Garg A, Strunk A, Midura M, Papagermanos V, Pomerantz H. Prevalence of hidradenitis suppurativa among patients with Down syndrome: a population-based cross-sectional analysis. *Br J Dermatol*. 2018;178(3):697–703
132. IRS. Able Act 2014. Available at: www.irs.gov/government-entities/federal-state-local-governments. Accessed August 7, 2020
133. White PH, Cooley WC; Transitions Clinical Report Authoring Group; American Academy of Pediatrics; American Academy of Family Physicians; American College of Physicians. Supporting the health care transition from adolescence to adulthood in the medical home. *Pediatrics*. 2018;142(5): e20182587
134. Wiseman FK, Al-Janabi T, Hardy J, et al. A genetic cause of Alzheimer disease: mechanistic insights from Down syndrome. *Nat Rev Neurosci*. 2015;16(9): 564–574
135. American College of Obstetricians and Gynecologists' Committee on Adolescent Health Care. Committee opinion no. 668: menstrual manipulation for adolescents with physical and developmental disabilities. *Obstet Gynecol*. 2016;128(2):e20–e25
136. Kirkham YA, Allen L, Kives S, Caccia N, Spitzer RF, Ornstein MP. Trends in menstrual concerns and suppression in adolescents with developmental disabilities. *J Adolesc Health*. 2013;53(3): 407–412
137. Got Transition. Available at: https://www.gottransition.org/six-core-elements/. Accessed March 25, 2022
138. Tsou AY, Bulova P, Capone G, et al. Global Down Syndrome Foundation Medical Care Guidelines for Adults with Down Syndrome Workgroup. Medical care of adults with Down syndrome: a clinical guideline. *JAMA*. 2020;324(15): 1543–1556
139. Rasmussen SA, Whitehead N, Collier SA, Frías JL. Setting a public health research agenda for Down syndrome: summary of a meeting sponsored by the Centers for Disease Control and Prevention and the National Down Syndrome Society. *Am J Med Genet A*. 2008;146A(23):2998–3010
140. Eunice Kennedy Shriver National Institute of Child Health and Human Development NIoH, Department of Health and Human Services. National Institutes of Health Research Plan on Down Syndrome (NA). Washington, DC: US Government Printing Office; 2007
141. National Institutes of Health. The INCLUDE Project. Available at: https:/www.nih.gov/include-project. Accessed March 25, 2022

# Supplemental Information

**SUPPLEMENTAL FIGURE 1.** Summary of Down syndrome-specific care.

| Action | Pre-natal | Birth up to 1 mo | 1 mo up to 1 yr | 1 yr up to 5 yr | 5 yr up to 12 yr | 12 yr up to 21 yr |
|---|---|---|---|---|---|---|
| 1. Confirm DS diagnosis with either CVS or amniocentesis prenatally or karyotype postnatally | | | | | | |
| 2. Review recurrence risk and offer the family referral to a clinical geneticist or genetic counselor. | | | | | | |
| 3. Offer parent-to-parent and support group information to the family. | | | | | | |
| 4. Use CDC DS-specific growth charts to monitor weight, length, weight-for-length, head circumference, or BMI. Use standard charts for BMI after age 10 years. | | All healthcare visits | | | | |
| 5. Order an echo, to be read by a pediatric cardiologist. | | | | | | |
| 6. Feeding assessment or video study if any: marked hypotonia, underweight (<5th %ile weight-for-length or BMI), slow feeding or choking with feeds, recurrent or persistent abnormal respiratory symptoms, desaturations with feeds | | Any visit | | | | |
| 7. Obtain objective hearing assessment (may be in NBS protocols) and follow EHDI protocols. | | | Up to 6 mo | | | |
| 8. If TM can't be visualized, refer to otolaryngologist for exam with microscope until reliable TM and tympanometry exams are possible | | Every 3-6 mo | | | | |
| 9. Car safety seat evaluation before hospital discharge. | | | | | | |
| 10. CBC with differential | | By day 3 | | | | |
| 11. If TAM, make caregivers aware of risk/signs of leukemia (e.g., easy bruising/bleeding, recurrent fevers, bone pain) | | | | | | |
| 12. TSH | | At birth (if not in NBS) | Every 5-7 mo | Annually, and every 6 mo if antithyroid antibodies ever detected | | |
| 13. RSV prophylaxis based on AAP guidelines. | | Annually | | Through 2 yr | | |
| 14. Discuss cervical spine-positioning for procedures and atlantoaxial stability precautions. | | All HMV | | Biennially | | |
| 15. Assess for CAM use, discourage any unsafe CAM practices. | | All HMV | | | | |
| 16. Refer children to early intervention for speech, fine motor or gross motor therapy. | | Any visit | Up to 3 yr | | | |
| 17. If middle ear disease occurs, obtain developmentally-appropriate hearing evaluation. | | | When ear clear | After treatment | | |
| 18. Rescreen hearing with developmentally-appropriate methodology (BAER, behavioral, ear-specific). | | | Start at 6mo, every 6 mo until established normal bilaterally by ear-specific testing, then annually | | | |
| 19. Refer to ophthalmologist with experience and expertise in children with disabilities. | | | By 6 mo | | | |
| 20. CBC with differential if easy bruising or bleeding, recurrent fevers, or bone pain | | | Any visit | | | |
| 21. Assess for sleep-disordered breathing; if present, refer to physician with expertise in pediatric sleep disorders. | | | At least once by 6 mo, then all subsequent HMV thereafter | | | |
| 22. Ensure child is receiving developmental therapies, and family understands and is following therapy plan at home. | | All HMV | | | | |
| 23. CBC with differential and either (1) a combination of ferritin and CRP, or (2) a combination of serum iron and Total Iron Binding Capacity | | | | Annually | | |
| 24. If a child has sleep problems and a ferritin less than 50 mcg/L, the pediatrician may prescribe iron supplement. | | | | Any visit | | |
| 25. Vision screening | | | All HMV, use developmentally-appropriate criteria | Photoscreen (all HMV); if unable, refer to ophthalmologist annually | Photoscreen (all HMV); if unable, refer to ophthalmologist biennially | Visual acuity or photoscreening at all HMV, or ophthalmology-determined schedule |
| 26. If a child has myelopathic symptoms, obtain neutral C-spine plain films (see text for details). | | | | Any visit | | |
| 27. Obtain polysomnogram. | | | | Between 3-5 yr | | |
| 28. Prepare family for transition from early intervention to preschool. | | | | At 30 mo | | |
| 29. Discuss sexual exploitation risks. | | | | At least once | At least once | At least once |
| 30. Make developmentally-appropriate plans for menarche, contraception (advocate/offer LARC), and STI prevention. | | | | | As developmentally-appropriate, then all subsequent HMV | |
| 31. Discuss risk of DS if patient were to become pregnant. | | | | | At least once | At least once |
| 32. Assess for any developmental regression. | | | All HMV | | | |
| 33. Discuss and facilitate transitions: education, work, finance, guardianship, medical care, independent living | | | | | All HMV starting at 10 yr | |

| | |
|---|---|
| | Do once at this age |
| | Do if not done previously |
| | Repeat at indicated intervals |
| (border) | See report for end point |

Abbreviations: DS, Down syndrome; CVS, Chorionic villus sampling; HMV, Health Maintenance Visit; BMI, Body mass index; CDC, Centers for Disease Control; EHDI, Early Hearing Detection and Intervention; NBS, Newborn screen; CAM, Complementary and alternative medicine; BAER, Brainstem auditory evoked response; TM, Tympanic membrane; TAM: transient abnormal myelopoiesis