Clinical Report—Health Supervision for Children With Down Syndrome

Marilyn J. Bull, MD, and the COMMITTEE ON GENETICS

abstract

These guidelines are designed to assist the pediatrician in caring for the child in whom a diagnosis of Down syndrome has been confirmed by chromosome analysis. 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 the genetic counseling provided. Therefore, this report offers guidance for this situation as well.

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INTRODUCTION

Children with Down syndrome have multiple malformations, medical conditions, and cognitive impairment because of the presence of extra genetic material from chromosome 21. 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, and short fifth finger with clinodactyly and wide spacing, often with a deep plantar groove between the first and second toes. The degree of cognitive impairment is variable and may be mild (IQ of 50–70), moderate (IQ of 35–50), or occasionally severe (IQ of 20–35). There is a significant risk of hearing loss (75%); obstructive sleep apnea (50%–79%); otitis media (50%–70%); eye disease (60%), including cataracts (15%); and severe refractive errors (50%); congenital heart defects (50%); neurologic dysfunction (1%–13%); gastrointestinal atresias (12%); hip dislocation (6%); thyroid disease (4%–18%); and, less commonly, transient myeloproliferative disorder (4%–10%) and later leukemia (1%) and Hirschsprung disease (<1%) (Table 1). The social quotient may be improved with early-intervention techniques, although the level of function is exceedingly variable. Children with Down syndrome often function more effectively in social situations than would be predicted on the basis of cognitive assessment results.

In approximately 95% of children with Down syndrome, the condition is sporadic because of nonfamilial trisomy 21, in which there are 47 chromosomes with a free extra chromosome 21 being present. In approximately 3% to 4% of persons 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. Approximately three-quarters of these

ABBREVIATIONS

BAER—brainstem auditory evoked response
TSH—thyroid-stimulating hormone
CRP—C-reactive protein

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The guidance in this report does not indicate an exclusive course of treatment or serve as a standard of medical care. Variations, taking into account individual circumstances, may be appropriate.
Physicians may be asked to counsel a family whose fetus has been identified with or is at increased risk of Down syndrome. In some settings, the pediatrician may be the primary resource for counseling. At other times, counseling may have been provided for the family by a certified genetic counselor, a clinical geneticist, obstetrician, or developmental-behavioral pediatrician. In addition, parents may have received information from a Down syndrome program, a national Down syndrome organization, or an Internet site. Because the pediatrician often has a previous relationship with the family, he or she should be prepared to review this information and assist in the decision-making process. When asked, the pediatrician should discuss the following topics with the family:

1. The prenatal laboratory studies that lead to the diagnosis and any fetal imaging studies that have been or will be performed.
2. The mechanism for occurrence of the disorder in the fetus and the potential recurrence rate for the family as provided by genetic counseling.
3. The prognosis and phenotypic manifestations, including the wide range of variability seen in infants and children with Down syndrome. Families benefit from hearing a fair and balanced perspective, including the many positive outcomes of Down syndrome and facilitate their transition to adulthood. The following outline is designed to help the pediatrician provide care for children with Down syndrome and their families in the medical home. It is organized by the issues that need to be addressed in various age groups (see Appendix 1).

Several areas require ongoing assessment throughout childhood and should be reviewed at every physician 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 for which the child and family may be eligible;
- injury and abuse prevention with special consideration of developmental skills; 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, be offered the option of diagnostic testing for Down syndrome and consider less invasive screening options. Screening options have improved significantly with the introduction of first-trimester screening, which incorporates maternal age, nuchal translucency ultrasonography, and measurement of maternal serum human chorionic gonadotropin (β-hCG) and pregnancy-associated plasma protein A (PAPP-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 screening, often called the quad screen, incorporates maternal age risk with measurement of maternal serum hCG, unconjugated estriol, α-fetoprotein (AFP), and inhibin levels. 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 approximately 95%. These screening tests are reported to have a 5% false-positive rate.

Medical management, home environment, early intervention, education, and vocational training can significantly affect the level of functioning of children and adolescents with Down syndrome and facilitate their transition to adulthood. The following outline is designed to help the pediatrician provide care for children with Down syndrome and their families in the medical home. It is organized by the issues that need to be addressed in various age groups (see Appendix 1). Several areas require ongoing assessment throughout childhood and should be reviewed at every physician visit and at least annually. These areas include:

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3. The prognosis and phenotypic manifestations, including the wide range of variability seen in infants and children with Down syndrome. Families benefit from hearing a fair and balanced perspective, including the many positive outcomes of

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### TABLE 1 Medical Problems Common in Down Syndrome

<table>
<thead>
<tr>
<th>Condition</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hearing problems</td>
<td>75</td>
</tr>
<tr>
<td>Vision problems</td>
<td>60</td>
</tr>
<tr>
<td>Cataracts</td>
<td>15</td>
</tr>
<tr>
<td>Refractive errors</td>
<td>50</td>
</tr>
<tr>
<td>Obstructive sleep apnea</td>
<td>50–75</td>
</tr>
<tr>
<td>Otitis media</td>
<td>50–70</td>
</tr>
<tr>
<td>Congenital heart disease</td>
<td>40–50</td>
</tr>
<tr>
<td>Hypodontia and delayed dental eruption</td>
<td>23</td>
</tr>
<tr>
<td>Gastrointestinal atresias</td>
<td>12</td>
</tr>
<tr>
<td>Thyroid disease</td>
<td>4–18</td>
</tr>
<tr>
<td>Seizures</td>
<td>1–13</td>
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<tr>
<td>Hematologic problems</td>
<td></td>
</tr>
<tr>
<td>Anemia</td>
<td>3</td>
</tr>
<tr>
<td>Iron deficiency</td>
<td>10</td>
</tr>
<tr>
<td>Transient myeloproliferative disorder</td>
<td>10</td>
</tr>
<tr>
<td>Leukemia</td>
<td>1</td>
</tr>
<tr>
<td>Celiac disease</td>
<td>5</td>
</tr>
<tr>
<td>Atlantoaxial instability</td>
<td>1–2</td>
</tr>
<tr>
<td>Autism</td>
<td>1</td>
</tr>
<tr>
<td>Hirschsprung disease</td>
<td>&lt;1</td>
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children with Down syndrome and their effect on the family.

4. Any additional studies performed that may refine the estimation of the prognosis (e.g., fetal echocardiogram, ultrasonographic examination for gastrointestinal tract malformations). Consultation with an appropriate medical subspecialist, such as a pediatric cardiologist or a pediatric surgeon, should occur prenatally if abnormal findings are detected.

5. Currently available treatments and interventions. This discussion needs to include the efficacy, potential complications and adverse effects, costs, and other burdens associated with treatments. Discuss early-intervention resources, parent support programs, and any appropriate future treatments.

6. The options available to the family for management and rearing of the child should be discussed using a nondirective approach. In cases of early prenatal diagnosis, this may include discussion of pregnancy continuation or termination, raising the child in the family, foster care placement, and adoption.

7. Availability of genetic counseling or meeting with a genetics professional.

If the pregnancy is continued:

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 subspecialty consultants, and/or if desired by the family for modifying this management plan (e.g., detection of a complex heart defect by echocardiography).

2. Offer parent-to-parent contact and information about local and national support organizations.

3. Offer referral to a clinical geneticist 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

Examination

The first step in evaluating a newborn infant for trisomy 21 is a careful review of the family history and prenatal information, particularly if prenatal chromosome studies were performed. Previous children born with trisomy 21 or developmental differences or pregnancies that ended in miscarriage 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 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, a blood sample should be obtained for postnatal cytogenetic analysis 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 feels that enough criteria are present on physical examination, then a blood sample should be sent for chromosome evaluation. The clinician should alert the laboratory and request rapid results. A study that uses fluorescent in situ hybridization (FISH) technology should be available within 24 to 48 hours to facilitate diagnosis and parent counseling. A FISH study can only indicate that an extra copy of chromosome 21 is present; it cannot detect translocations. Therefore, a positive FISH-test result should be confirmed by a complete chromosome analysis to identify translocations that may have implications for further reproductive counseling for the parents and possibly other family members.

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. It is recommended that hospitals coordinate the delivery of the information and offer a private hospital room pending confirmation of the diagnosis.

An important aspect of providing information about Down syndrome to families includes first congratulating 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, where appropriate, with both parents together. Physicians should use their experience and expertise in providing support and guidance for families. Clinicians should ensure a balanced approach rather than their personal opinions, 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 healthy, productive lives of people with Down syndrome in society.

Confirm the laboratory diagnosis of Down syndrome and review the karyotype with the parents when the final result is available. Discuss the specific findings with both parents whenever possible, and talk about the potential clinical manifestations associated with the syndrome. These topics...
should be reviewed again at a subsequent meeting. Parents should be referred 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 provider, and extreme care is required to be certain that a smooth transition occurs for the family.

Discuss and Review

- Hypotonia.
- Facial appearance, and acknowledge the presence of familial characteristics.
- Feeding issues. Children with Down syndrome can usually nurse, and many can breastfeed successfully. Occasionally, some will need early supplementation until a successful nursing pattern is established. Some infants will also sleep for prolonged periods and need to be awakened to feed to maintain adequate calorie intake.

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. Refer all infants who have marked hypotonia as well as infants with slow feeding, choking with feeds, recurrent pneumonia, or other recurrent or persistent respiratory symptoms and unexplained failure to thrive for a radiographic swallowing assessment.\(^{14,15}\)
- Cataracts at birth by looking for a red reflex. Cataracts may progress slowly and, if detected, need prompt evaluation and treatment by an ophthalmologist with experience in managing the child with Down syndrome.
- Congenital hearing loss, with objective testing, such as brainstem auditory evoked response or otoacoustic emission, at birth, according to the universal newborn hearing screening guidelines. Complete any needed follow-up assessment by 3 months.\(^{16,17}\)
- Duodenal atresia or anorectal atresia/stenosis by performing a history and clinical examination.
- Apnea, bradycardia, or oxygen desaturation in a car safety seat for infants who are at increased risk because they have had cardiac surgery or are hypotonic. A car safety seat evaluation should be conducted for these infants before hospital discharge.\(^{18}\)
- Constipation. If constipation is present, evaluate for restricted diet or limited fluid intake, hypotonia, hypothyroidism, or gastrointestinal tract malformation, including stenoses or Hirschsprung disease, for which there is an increased risk.
- Gastroesophageal reflux, which is usually diagnosed and managed clinically. If severe or contributing to cardiorespiratory problems or failure to thrive, refer for subspecialty intervention.
- Stridor, wheezing, or noisy breathing. If severe or contributing to cardiorespiratory problems or feeding difficulty, refer to pediatric pulmonologist to assess for airway anomalies. Tracheal anomalies and small tracheal size may also make intubation more difficult.
- Hematologic abnormalities. Obtain a complete blood cell count. Leukemoid reaction, or transient myeloproliferative disorder (TMD). TMD is found almost exclusively in newborn infants with Down syndrome and is relatively common in this population (10%).\(^{19}\)

Anticipatory Guidance Given at Least Once Between Birth and 1 Month of Age

- 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, aggressive treatment should be instituted.\(^{14}\) Children with comorbid conditions who qualify should have respiratory syncytial virus prophylaxis.\(^{22}\)
Discuss with parents the importance of cervical spine-positioning precautions to avoid excessive extension or flexion to protect the cervical spine during any anesthetic, surgical, or radiographic procedure.23,24

Discuss efficacy of early intervention and availability of early-intervention services and therapies in the community. Initiate referral as appropriate.25

Inform the family of the availability of support and advice from parents of other children with Down syndrome.

Supply names of Down syndrome support groups and current books and pamphlets (see “Resources for Parents”).

Discuss the strengths of the child and positive family experiences.

Discuss the individual resources for support, such as family, clergy, and friends.

Talk about how and what to tell siblings, other family members, and friends. Review methods of coping with long-term disabilities.

Review the recurrence risk in subsequent pregnancies and the availability of prenatal diagnosis as provided in genetic counseling.

Discuss treatments that are considered complementary and alternative. Parents need an opportunity to learn objectively which therapies are safe and which are potentially dangerous (eg, cell therapy that may transmit slow viruses and fat-soluble vitamins that can cause toxicity). Several articles and Internet sites evaluate the legitimacy of claims that are made.26–28

Renal and urinary tract anomalies have been reported to occur at increased frequency among persons with Down syndrome, and screening for these anomalies for all children with Down syndrome has been suggested.29 Until studies confirm this finding and document that screening improves outcomes, routine renal and urologic screening is not recommended.

HEALTH SUPERVISION FROM 1 MONTH TO 1 YEAR: INFANCY

Physical Examination and Laboratory Studies

Review the risk of serous otitis media (50%–70%). Review the previous hearing evaluation (brainstem auditory evoked response [BAER, ABR] or otoacoustic emission). If the child passed the screening study, rescreen at 6 months of age for confirmation. If the infant failed to pass screening studies, refer to an otolaryngologist who is comfortable with examining infants with stenotic external canals to determine if a middle-ear abnormality is present. Tympanometry may be necessary if the tympanic membrane is poorly visualized. Middle-ear disease should be treated promptly. Once 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 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. A behavioral audiogram may be attempted at 1 year of age, but many children will not be able to complete the study and may need additional testing by BAER.30,31

At least once during the first 6 months of life, discuss with parents 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 above-mentioned symptoms occur.32,33

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. Perform careful history and physical examination, and pay attention for myelopathic signs and symptoms.

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, and nystagmus.34 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.35

Verify results of newborn thyroid-function screen if not previously performed. Because of increased risk of acquired thyroid disease, repeat measurement of TSH at 6 and 12 months of age and then annually.

Monitor infants with cardiac defects, 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
Anticipatory Guidance

- Monitor weight and follow weight-for-height trends at each health care visit. Review the infant’s growth and plot it by using the standard growth charts of the National Center for Health Statistics or the World Health Organization. The previously used Down syndrome-specific growth charts no longer reflect the current population styles and body proportion. Until new charts are developed, patterns of growth and weight gain should be followed on the available standard growth charts and should include use of weight for height and BMI.

- Review availability of Down syndrome support groups at least once in the first year of life (see “Resources for Parents”).

- Assess the emotional status of parents and intrafamilial relationships at each well-child visit. Educate and support siblings and discuss sibling adjustments.

- Review connection to early intervention services and their relationship to the strengths and needs of the infant and family at each well-child visit.

- Review the family’s understanding of the risk of recurrence of Down syndrome and the availability of prenatal diagnosis at least once in the first year of life and more often if judged necessary by the clinician. Refer for genetic counseling if not already provided.

- 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

- Obtain a history and perform a physical examination, and give attention to growth and developmental status at every well-child visit.

- Review the risk of hearing loss associated with serous otitis media. For a child who passed diagnostic hearing testing, additional screening or 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 a hearing loss should be referred to an otolaryngologist who is comfortable with the examination of children with stenotic ear canals. The risk of serious otitis media between 3 and 5 years of age is approximately 50% to 70%.

- Check the child’s vision, and use developmentally appropriate subjective and objective criteria at each well-child visit. Refer the child annually 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.\textsuperscript{34,49–51}

**Atlantoaxial Instability**

Discuss with parents, at least biennially, the importance of cervical spine positioning 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 Asymptomatic Child**

Children with Down syndrome are at increased risk of atlantoaxial subluxation. However, the child must be 3 years of age to have adequate vertebral mineralization and epiphyseal development for accurate radiographic evaluation of the cervical spine.\textsuperscript{52} 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.\textsuperscript{53,54} 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.\textsuperscript{55–64} Parents should be advised that participation in some sports, including contact sports such as football and soccer and gymnastics (usually at older ages), places children at increased risk of spinal cord injury\textsuperscript{65} and that trampoline use should be avoided by all children with or without Down syndrome younger than 6 years and by older children unless under direct professional supervision.\textsuperscript{66,67} Special Olympics has specific screening requirements for participation in some sports.\textsuperscript{68}

**The Symptomatic Child**

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.\textsuperscript{55,65} 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 in the neutral position, referral to a pediatric neurosurgeon or pediatric orthopedic surgeon 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 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 in the neutral position, further radiographs may be obtained before the patient is promptly referred.\textsuperscript{23,62,63}

- Measure TSH annually or sooner if child has symptoms that could be related to thyroid dysfunction.
- For children on a diet that contains gluten, at each preventative care visit review for symptoms potentially related to celiac disease, including diarrhea or protracted constipation, slow growth, unexplained failure to thrive, anemia, abdominal pain or bloating, or refractory developmental or behavioral problems.\textsuperscript{69–71} For those with symptoms, obtain a tissue transglutaminase immunoglobulin A (IgA) level and simultaneous quantitative IgA. The quantitative IgA is important, because a low IgA level will result in a false-negative tissue transglutaminase IgA result. Refer patients with abnormal laboratory values for specialty assessment. There is no evidence showing routine screening of asymptomatic individuals as being 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 obstructive sleep apnea, including heavy breathing, snoring, restless sleep, uncommon sleep positions, frequent night awakening, daytime sleepiness, apneic pauses, and behavioral problems, that could be associated with poor sleep at each well-child visit. There is poor correlation between parent report and polysomnogram results.\textsuperscript{35,72} Therefore, referral to a pediatric sleep laboratory for a sleep study or polysomnogram for all children with Down syndrome by 4 years of age 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. Discuss obesity as a risk factor for sleep apnea.\textsuperscript{34} It is recognized that access to a pediatric sleep laboratory or specialist may be limited for some populations and geographic areas.

- Maintain follow-up with a pediatric cardiologist for patients with cardiac lesions even after complete repair to monitor for recurrent/residual lesions as well as development of pulmonary hypertension.

- Monitor for neurologic dysfunction, including seizures.

- Obtain hemoglobin concentration

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annually. Also, obtain serum ferritin and CRP concentrations for any child at risk of iron deficiency.

**Anticipatory Guidance**

- Review early intervention, including physical therapy, occupational therapy, and speech therapy, at all health maintenance 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 (IFSP) in early intervention to the Individualized Education Plan (IEP) through public education.

- Discuss with caregivers at every visit the child’s behavioral and social progress. Refer children who may have autism, attention-deficit/hyperactivity disorder, or other psychiatric or behavioral problems for appropriate evaluation and intervention as soon as suspected. 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.22

- Provide influenza vaccine annually. Children with chronic cardiac or pulmonary disease should be given the 23-valent pneumococcal polysaccharide vaccine (PPS23) at 2 years or older.22

- Reassure parents that delayed and irregular dental eruption patterns are common and that hypodontia occurs with increased frequency (23%).77,78

- Encourage and model use of accurate terms for genitalia and other private body parts (penis, vulva) any time these body parts are discussed or examined. Model respect for body rights by reminding patients that their body is their own and explain to the child what you will do before moving into child’s 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).79

- On at least 1 well-child visit educate parents about increased risk of sexual exploitation, and remind them that likely perpetrators are people their child knows and trusts, not strangers.

- At least once between 1 and 5 years of age, as with discussion in the first year of life, discuss future pregnancy planning and review risk of recurrence of Down syndrome and availability of prenatal diagnosis.

- Assess the child’s behavior and talk about behavioral management, sibling adjustments, socialization, and recreational skills.

- 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 and alternative.

**HEALTH SUPERVISION FROM 5 TO 13 YEARS: LATE CHILDHOOD**

- Obtain a history and perform a physical examination with attention to growth and developmental status at each annual well-child visit.

- Monitor growth patterns, especially BMI, and emphasize healthy diet and lifestyle for preventing obesity.

- Obtain annual ear-specific audiologic evaluation.

- Obtain ophthalmologic evaluation every 2 years.

- Measure TSH annually; the risk of hypothyroidism increases with age.

- Individualize cardiology follow-up on the basis of history of cardiac defects.

- Obtain hemoglobin concentration annually and serum ferritin and CRP or reticulocyte hemoglobin concentrations at annual visits for any child at risk of iron deficiency on the basis of history of decreased iron intake.

- 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.

- At each well-child visit, discuss with parents 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. Parents should also be instructed to contact their physician immediately for new onset of symptoms of myelopathy.

- Counsel parents that some sports place children at increased risk of spinal cord injury.65–67

- Monitor for neurologic dysfunction, including seizures.

- Very dry skin, which may be a sign of hypothyroidism, and other skin problems are particularly common in patients with Down syndrome. Therefore, be attentive to these dermatologic problems and discuss them with the patient and family.

- Discuss symptoms related to obstructive sleep apnea 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 any child with signs or symptoms of obstructive sleep apnea or abnormal sleep-
study results. Discuss obesity as a risk factor of sleep apnea.

Anticipatory Guidance at Every Health Maintenance Visit

- Review the child’s development and appropriateness of school placement and developmental intervention.
- Discuss socialization, family status, and relationships, including financial arrangements, health insurance, and guardianship.
- Discuss the development of age-appropriate social skills, self-help skills, and development of a sense of responsibility.
- 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 seen in typically developing children may also occur. Evaluate for medical problems that can be associated with behavior changes, including thyroid abnormalities, celiac disease, sleep apnea, 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. Referral to community treatment programs, psychosocial services for consultative care, or behavioral specialists experienced in working with children with special needs may be necessary. The use of medication for behavior management should be discussed between the primary care physician and specialists involved in the child’s care, because children with Down syndrome may be more sensitive to certain medications. Although there has been little research to directly address the use of psychotropic medications among children with Down syndrome, anecdotal reports indicate that such children may differ in their response to medications.
- Counsel families regarding the transition from elementary to middle school, when major change often occurs, from 1 to many teachers 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.
- Refer children who may have autism for appropriate evaluation and intervention as soon as suspected.
- Continue to assess, monitor, and encourage independence with hygiene and self-care. Encourage parents 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. Remind parents that physical 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 them.
- Discuss the need for gynecologic care in the pubescent girl. Talk with the patient and her family about the recurrence risk of Down syndrome (50%) were she to become pregnant. Although males with Down syndrome are usually infertile, there have been rare instances in which a male has reproduced. Birth control and prevention of sexually transmitted diseases should be discussed with patients and their families. Families may wish to discuss sterilization, and the pediatrician may review the topic in the American Academy of Pediatrics policy statement “Sterilization of Minors With Developmental Disabilities.”
- Be prepared to discuss and answer questions regarding treatments that are considered complementary and alternative.

HEALTH SUPERVISION FROM 13 TO 21 YEARS OR OLDER: ADOLESCENCE TO EARLY ADULTHOOD

Physical Examination and Laboratory Values

- Measure hemoglobin concentration annually.
- Measure TSH concentration annually.
- Obtain annual ear-specific audiologic evaluation.
- 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.
- Individualize cardiology follow-up on the basis of history of cardiac defects. Discuss symptoms related to obstructive sleep apnea, including snoring, restless sleep, daytime sleepiness, nighttime awakening, behavior problems, and sleep position at every health maintenance visit. Refer to a physician with expertise in pediatric sleep any child with signs or symptoms of obstructive sleep apnea or an abnormal sleep-study result. Discuss the risk factor of obesity for sleep apnea.
- Discuss with parents and the patient at every 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 myopathic signs and symptoms. Parents and patients should also be instructed to contact their physician immediately for new onset of symptoms of myopathy.

- Counsel parents that some sports place children at increased risk of spinal cord injury.85-87
- Monitor for signs of other neurologic dysfunction, including seizures.
- Obtain ophthalmologic evaluation every 3 years. Check for onset of cataracts, refractive errors, and keratoconus, which can cause blurred vision, corneal thinning, or corneal haze and is typically diagnosed after puberty.
- 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, or exertional dyspnea or abnormal physical examination findings, such as a new murmur or gallop. Discuss skin, hair, and scalp care at each preventive health care visit.

**Anticipatory Guidance at Every Health Maintenance Visit**

- Discuss issues related to transition into adulthood, including guardianship and long-term financial planning from early adolescence. Potential adult morbidities including apparent tendency toward premature aging and increased risk of Alzheimer disease may also be discussed.87
- Monitor growth patterns, especially BMI, and counsel regarding healthy diet and a structured exercise program.
- Discuss behavioral and social states and refer patients who have chronic behavioral problems or manifest acute deterioration in function for specialized evaluation and intervention.88,89
- Discuss appropriateness of school placement, and emphasize planning for transition to adulthood and adequate vocational training within the school curriculum.90,91
- Talk with the female patient and her family about the recurrence risk of Down syndrome should she become pregnant.
- Continue to assess, monitor, and encourage independence with hygiene and self-care. Provide guidance on healthy, normal, and typical sexual development and behaviors. Emphasize the need for understandable information, and encourage opportunities for advancing comprehension of sexuality. Discuss the need for contraception and prevention of sexually transmitted diseases and the degree of supervision required. Advocate for the least invasive and least permanent method of birth control and be familiar with local law and resources to assist the family in their decision-making regarding questions about sterilization.96
- Make recommendations and provide or refer for routine gynecologic care if not already provided. Discuss premenstrual behavioral problems and management of menses.92
- Discuss group homes and independent living opportunities, workshop settings, and other community-supported employment.
- Discuss intrafamily relationships, financial planning, and guardianship.
- Facilitate transition to adult medical care.93

**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 for atlantoaxial instability, for example, is needed,94 and continuing research is critical for directing the care for optimal outcomes of persons with Down syndrome.1,95,96

**ACKNOWLEDGMENT**

The mentoring and contributions of Dr. William Cohen have been sincerely appreciated and were integral to the development of this clinical report. His untimely death is a great loss to his patients and their families, his colleagues, and the greater medical community. This clinical report is dedicated to his memory.

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**RESOURCES FOR PARENTS**

REFERENCES


29. Kupferman J, Druschel C, Kuchich G. Increased prevalence of renal and urinary tract anomalies in children with Down syn-


## APPENDIX 1  Health Supervision for Children With Down Syndrome

<table>
<thead>
<tr>
<th>Visit Category</th>
<th>Prenatal</th>
<th>Birth-1 mo</th>
<th>1 mo-1 y</th>
<th>1-5 y</th>
<th>5-13 y</th>
<th>13-21 y</th>
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</thead>
<tbody>
<tr>
<td>Counseling regarding prenatal screening test &amp; imaging results</td>
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<td>Plan for delivery</td>
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<td>Referral to geneticist</td>
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<td>Parent-to-parent contact, support groups, current books and pamphlets</td>
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<td>Physical exam for evidence of trisomy 21</td>
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<td>Chromosomal analysis to confirm dx</td>
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<td>Discuss risk of recurrence of Down syndrome</td>
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<td>Ophthalmogram</td>
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<td>Radiographic swallowing assessment if marked hypotonia, slow feeding, choking with feeds, recurrent or persistent respiratory sx, FTT</td>
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<td>Thy and FE assessment for duodenal or anorectal atresia</td>
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<td>Reassure parents delayed and irregular dental eruption, hypodontia are common</td>
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<td>If cardiomyopathy, evaluate for limited fed or fluids, hypoxemia, neurohypothyroidism, GI malformation, Hirschsprung</td>
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<td>CBC to R/O transient mycoproliferative disorder, polycythemia</td>
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<td>Hb annually; CRP &amp; ferritin or CHr if possible risk iron deficiency or Hb &lt;12 g.</td>
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<td>Hemoglobin</td>
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<tr>
<td>LDH (may be part of newborn screening)</td>
<td>6 and 12 mo</td>
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<td>Discuss risk of respiratory infection</td>
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<td>If cardiac surgery or hypertonic: evaluate apex, bradycardia, or oxygen desaturation in car seat before discharge</td>
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<td>Discuss complementary &amp; alternative therapies</td>
<td>All health maint. visits</td>
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<td>Discuss cervical spine positioning, especially for anesthesia or surgical or radiologic procedure</td>
<td>All health maint. visits</td>
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<tr>
<td>Review signs and symptoms of myopathy</td>
<td>All health maint. visits</td>
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<tr>
<td>If myopathic signs or symptoms: obtain neutral position spine films and, if normal, obtain flexion &amp; extension films &amp; refer to pediatric neurosurgeon or orthopedic surgeon with expertise in evaluating and treating atlanto-axial instability</td>
<td>Any visit</td>
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<tr>
<td>Institute to contact physician for change in gait, change in use of arms or hands, change in bowel or bladder function, neck pain, head tilt, torticollis, or new-onset weakness</td>
<td>Biennially</td>
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<td>Advise risk of some contact sports, trampolines</td>
<td>All health maint. visits</td>
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<td>Audiology evaluation at 18 mo</td>
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<tr>
<td>If normal hearing established, behavioral audiogram and tympanometry until bilateral ear specific testing possible. Refer child with abnormal hearing to ot</td>
<td>Every 6 mo</td>
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<tr>
<td>If normal ear-specific hearing established, behavioral audiogram</td>
<td>Annually</td>
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<td>Assess for obstructive sleep apnea 1 y</td>
<td>All health maint. visits</td>
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<td>Sleep study by age 3 y</td>
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<td>Ophthalmology referral to assess for strabismus, cataracts, and nystagmus</td>
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<td>Refer to pediatric ophthalmologist or ophthalmologist with experience with Down syndrome</td>
<td>Annually</td>
<td>Every 2 y</td>
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<tr>
<td>If congenital heart disease, monitor for signs &amp; sx of Congestive heart failure</td>
<td>All visits</td>
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<td>Assess the emotional status of parents and intrafamilial relationships</td>
<td>All health maint. visits</td>
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<td>Check for 5c of celiac disease; If 5c present, obtain tissue transglutaminase IgA &amp; quantitative lgA</td>
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<td>Early intervention: physical, occupational, and speech therapy</td>
<td>All health maint. visits</td>
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<td>At 30 months, discuss transition to preschool and development of IEP</td>
<td>All health maint. visits</td>
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<td>Discuss behavioral and social progress</td>
<td>Health maint. visits</td>
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<tr>
<td>Discuss self-help skills, ADHD, OCD, wandering off, transition to middle school</td>
<td>Health maint. visits</td>
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<td>If chronic cardiac or pulmonary disease, 23-valent pneumococcal vaccine at age &gt;2 y</td>
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<td>Assess regarding delayed and irregular dental eruption</td>
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<tr>
<td>Establish optimal dietary and physical exercise patterns</td>
<td>Health maint. visits</td>
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<td>Discuss dermatologic issues with parents</td>
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<td>Discuss physical and psychosocial changes through puberty; need for gynecologic care in the pubescent female</td>
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<td>Facilitate transition: guardianship, financial planning, behavioral problems, school placement, vocational training, independence with hygiene and self-care, group homes, work settings</td>
<td>Health maint. visits</td>
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<tr>
<td>Discuss sexual development and behaviors, contraception, sexually transmitted diseases, recurrence risk for offspring</td>
<td>Health maint. visits</td>
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</tbody>
</table>

Maint. indicates maintenance; dx, diagnosis; sx, symptoms; FTT, failure to thrive; Hx, history; FE, physician examination; GI, gastrointestinal; CBC, complete blood count; R/O, rule out; Hb, hemoglobin; ot, occupational therapy; CHr, reticuloocyte hemoglobin; lgA, immunoglobulin A; IEP, Individualized Education Plan; ADHD, attention-deficit/hyperactivity disorder; OCD, obsessive compulsive disorder.
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