

# AMERICAN ACADEMY OF ACADEMY OF PEDIATRICS

Committee on Genetics

## Health Supervision for Children With Down Syndrome

**ABSTRACT.** These guidelines are designed to assist the pediatrician in caring for the child in whom the diagnosis of Down syndrome has been confirmed by karyotype. Although the pediatrician's initial contact with the child is usually during infancy, occasionally the pregnant woman who has been given the prenatal diagnosis of Down syndrome will be referred for counseling. Therefore, these guidelines offer advice for this situation as well.

Children with Down syndrome have multiple malformations and mental impairment because of the presence of extra genetic material from chromosome 21. Although the phenotype is variable, usually there is enough consistency to enable the experienced clinician to suspect the diagnosis. Among the more common physical features are hypotonia, small brachycephalic head, epicanthic 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. A wide space, often with a deep fissure between the first and second toes, is also common. The degree of mental impairment is variable, ranging from mild (IQ: 50–70) to moderate (IQ: 35–50), and only occasionally to severe (IQ: 20–35). There is an increased risk of congenital heart defects (50%); leukemia (<1%); hearing loss (75%); otitis media (50%–70%); Hirschsprung disease (<1%); gastrointestinal atresias (12%); eye disease (60%), including cataracts (15%) and severe refractive errors (50%); acquired hip dislocation (6%); obstructive sleep apnea (50%–75%); and thyroid disease (15%). The social quotient may be improved with early intervention techniques, although the level of function is exceedingly variable. Children with Down syndrome often function better in social situations than might be expected from their IQ.

In approximately 95% of children with Down syndrome, the condition is because of nonfamilial trisomy 21. In approximately 3% to 4% of persons with the Down syndrome phenotype, the extra chromo-

somal material is the result of an unbalanced translocation between chromosome 21 and another acrocentric chromosome, usually chromosome 14. Approximately three fourths of these unbalanced translocations are de novo, and approximately one fourth are the result of familial translocations. If the child has a translocation, a balanced translocation must be excluded in the parents. If there is a translocation in either parent, additional familial studies and counseling should be instituted. In the remaining 1% to 2% of persons with the Down syndrome phenotype, 2 cell lines are present: 1 normal and 1 trisomy 21. This condition is called mosaicism. These persons, on average, may be phenotypically less severely affected than persons with trisomy 21 or translocated chromosome 21, but their conditions are generally indistinguishable in all other aspects.

Medical management, home environment, 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 to care for children with Down syndrome and their families.<sup>1–4</sup> It is organized by the issues that need to be addressed in the various age groups (see Table 1).

Several areas require ongoing assessment throughout childhood and should be reviewed periodically at developmentally appropriate ages. These include the following:

- Personal support available to family.
- All other financial and medical support programs for which the child and family may be eligible.
- Supplemental Security Income benefits.
- Injury and abuse prevention with special consideration of developmental skills.
- Diet and exercise to maintain appropriate weight.

### THE PRENATAL VISIT

Pediatricians may be asked to counsel a family in which a fetus has a genetic disorder. 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 clinical geneticist, obstetrician, or developmental pediatrician. In addition, parents may have received information from a Down syndrome program, a national Down syndrome organization, or an Internet site. Because of a previous relationship with the family, the pediatrician may be asked to review this information and assist in the decision-making process. As appropriate, the pediatrician should discuss the following topics with the family:

The recommendations in this policy statement do not indicate an exclusive course of treatment for children with genetic disorders, but are meant to supplement anticipatory guidelines available for treating the healthy child provided in the AAP publication, "Guidelines for Health Supervision." They are intended to assist the pediatrician in helping children with genetic conditions to participate fully in life. Diagnosis and treatment of genetic disorders are changing rapidly. Therefore, pediatricians are encouraged to view these guidelines in the light of evolving scientific information. Clinical geneticists may be a valuable resource for the pediatrician seeking additional information or consultation.

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TABLE 1. Health Supervision for Children With Down Syndrome—Committee on Genetics\*

|                              | Prenatal |          | Infancy, 1 Month to 1 Year |          |          |           |           | Early Childhood, 1 to 5 Years |           |         |         |                       | Late Childhood, Adolescence, 5 to 13 Years, 13 to 21 Years, Annual |  |
|------------------------------|----------|----------|----------------------------|----------|----------|-----------|-----------|-------------------------------|-----------|---------|---------|-----------------------|--|--|
|                              | Neonatal | 2 Months | 4 Months                   | 6 Months | 9 Months | 12 Months | 15 Months | 18 Months                     | 24 Months | 3 Years | 4 Years | 5 to 13 Years, Annual | 13 to 21 Years, Annual   |  |
| Diagnosis                    |          |          |                            |          |          |           |           |                               |           |         |         |                       |  |  |
| Karyotype review†            | •        |          |                            |          |          |           |           |                               |           |         |         |                       |  |  |
| Phenotype review             | •        |          |                            |          |          |           |           |                               |           |         |         |                       |  |  |
| Recurrence risks             | •        |          |                            |          |          |           |           |                               |           |         |         |                       |  |  |
| Anticipatory guidance        |          |          |                            |          |          |           |           |                               |           |         |         |                       |  |  |
| Early intervention services  | •        | •        | •                          | •        | •        | •         | •         | •                             | •         | •       | •       | •                     | •  |  |
| Reproductive options         | •†       | •†       | •                          | •        | •        | •         | •         | •                             | •         | •       | •       | •                     | •  |  |
| Family support               | •        | •        | •                          | •        | •        | •         | •         | •                             | •         | •       | •       | •                     | •  |  |
| Support groups               | •        | •        | •                          | •        | •        | •         | •         | •                             | •         | •       | •       | •                     | •  |  |
| Long-term planning           | •        |          |                            |          |          |           |           |                               |           |         |         |                       |  |  |
| Sexuality                    |          |          |                            |          |          |           |           |                               |           |         |         |                       |  |  |
| Medical evaluation           |          |          |                            |          |          |           |           |                               |           |         |         |                       |  |  |
| Growth                       | o        | o        | o                          | o        | o        | o         | o         | o                             | o         | o       | o       | o                     | o  |  |
| Thyroid screening            | o        |          |                            |          |          |           |           |                               |           |         |         |                       |  |  |
| Hearing screening            | o        | S/o      | S/o                        | S/o      | S/o      | S/o†      | S/o       | S/o                           | S/o       | S/o     | S/o     | S/o                   | S/o  |  |
| Vision screening             | S/o      | S/o†     | S/o                        | S/o      | S/o†     | S/o       | S/o       | S/o                           | S/o       | S/o     | S/o     | S/o                   | S/o  |  |
| Cervical spine roentgenogram |          |          |                            |          |          |           |           |                               |           |         |         |                       |  |  |
| Echocardiogram               | •        |          |                            |          |          |           |           |                               |           |         |         |                       |  |  |
| CBC                          | o        |          |                            |          |          |           |           |                               |           |         |         |                       |  |  |
| Psychosocial                 |          |          |                            |          |          |           |           |                               |           |         |         |                       |  |  |
| Development and behavioral   | S/o      | S/o      | S/o                        | S/o      | S/o      | S/o       | S/o       | S/o                           | S/o       | S/o     | S/o     | S/o                   | S/o  |  |
| School performance           |          |          |                            |          |          |           |           |                               |           |         |         |                       |  |  |
| Socialization                | S        |          |                            |          |          |           |           |                               |           |         |         |                       |  |  |

\* Assure compliance with the American Academy of Pediatrics "Recommendations for Preventive Pediatric Health Care."

• = to be performed; S = subjective, by history; and o = objective, by a standard testing method.

† Or at time of diagnosis.

‡ Discuss referral to specialist.

§ Give once in this age group.

|| According to state law.

|| As needed.

\*\* See discussion.

1. The prenatal laboratory or fetal imaging studies leading to the diagnosis.
2. The mechanism for occurrence of the disorder in the fetus and the potential recurrence rate for the family.
3. The prognosis and manifestations, including the wide range of variability seen in infants and children with Down syndrome.
4. When applicable, additional studies that may refine the estimation of the prognosis (eg, fetal echocardiogram, ultrasound examination for gastrointestinal malformations).
5. Currently available treatments and interventions. This discussion needs to include the efficacy, potential complications and adverse effects, costs, and other burdens associated with these treatments. Discuss early intervention resources, parent support programs, and any plausible future treatments.
6. The options available to the family for management and rearing of the child using a nondirective approach. In cases of early prenatal diagnosis, this may include discussion of pregnancy continuation or termination, rearing the child at home, foster care placement, and adoption.

If the pregnancy is continued, a plan for delivery and neonatal care must be developed with the obstetrician and the family. Offer parent-to-parent contact. As the pregnancy progresses, additional studies may be valuable for modifying this management plan (eg, detection of a complex heart defect by echocardiography). When appropriate, referral to a clinical geneticist should be considered 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: NEWBORNS

##### Examination

Confirm the diagnosis of Down syndrome and review the karyotype with the parents. Review the phenotype. Discuss the specific findings with both parents whenever possible, and talk about the following potential clinical manifestations associated with the syndrome. These may have to be reviewed again at a subsequent meeting.

##### *Discuss and Review*

- Hypotonia
- Facial appearance

##### *Evaluate for*

- Feeding problems
- Strabismus, cataracts, and nystagmus at birth or by 6 months
- Congenital hearing loss with objective testing, such as brainstem auditory evoked response or otoacoustic emission at birth or by 3 months<sup>5</sup>
- Heart defects (approximately 50% risk). Perform cardiac evaluation (consultation by a cardiologist

with expertise and experience in pediatric patients and echocardiogram recommended).

- Duodenal atresia
- Constipation with increased risk of Hirschsprung disease
- Leukemia—more common in children with Down syndrome than in the general population, but still rare (<1%); leukemoid reactions, on the other hand, are relatively common as is polycythemia (18%). Obtain complete blood count.
- Congenital hypothyroidism (1% risk)
- Respiratory tract infections

##### *Anticipatory Guidance*

- Discuss increased susceptibility to respiratory tract infection
- Discuss the availability and efficacy of early intervention.
- Discuss the early intervention services in the community.
- Inform the family of the availability of support and advice from the parents of other children with Down syndrome.
- Supply names of Down syndrome support groups and current books and pamphlets (see “Bibliography and Resources for New Parents”).
- Discuss the strengths of the child and positive family experiences.
- Check on individual resources for support, such as family, clergy, and friends.
- Talk about how and what to tell 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.
- Discuss unproven therapies.<sup>6–12</sup>

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

##### Examination

##### *Physical Examination and Laboratory Studies*

- Review the risk of serous otitis media (50%–70%). If the tympanic membranes cannot be visualized, or if the parents express any concern about their child’s hearing, refer the infant to an otolaryngologist. Review the prior hearing evaluation (brainstem auditory evoked response and otoacoustic emission and refer back to the otolaryngologist and audiologist if the initial evaluation was abnormal for follow up examination and testing. A behavioral audiogram should be obtained at 1 year in all children examined.
- Check for strabismus, cataracts, and nystagmus by 6 months, if not done at birth. Check the infant’s vision at each visit, using developmentally appropriate subjective and objective criteria. By 6 months, refer the infant to a pediatric ophthalmologist or an ophthalmologist with special expertise and experience with infants with disabilities.
- Verify results of newborn thyroid function screen. Because of increased risk of acquired thyroid disease, repeat at 6 and 12 months and then annually.<sup>13,14</sup>

- Administer pneumococcal vaccine, as well as other vaccines recommended for all children unless there are specific contraindications.

**Anticipatory Guidance**

- Review the infant’s growth and development relative to other children with Down syndrome (Figs 1–4).<sup>15</sup>
- Review availability of Down syndrome support groups (see “Bibliography and Resources for New Parents”).
- Assess the emotional status of parents and intrafamily relationships. Educate and support siblings and discuss sibling adjustments. At 6 to 12 months, review the psychological support and intrafamily relationships, including long-term planning, financial planning, and guardianship.
- Review the early intervention services relative to the strengths and needs of the infant and family

(see “Bibliography and Resources for New Parents”).

- Review the family’s understanding of the risk of recurrence of Down syndrome and the availability of prenatal diagnosis.

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

- Obtain a history and perform a physical examination with attention to growth and developmental status.
- Review the risk of serous otitis media with hearing loss. If the tympanic membranes cannot be completely visualized (because of the frequent problem of stenotic ear canals), check the child’s audiogram every 6 months up to 3 years or up to when a pure tone audiogram is obtained. Refer the child to an otolaryngologist or audiologist if necessary

**DOWN SYNDROME GIRLS  
PHYSICAL GROWTH:  
1 TO 36 MONTHS**

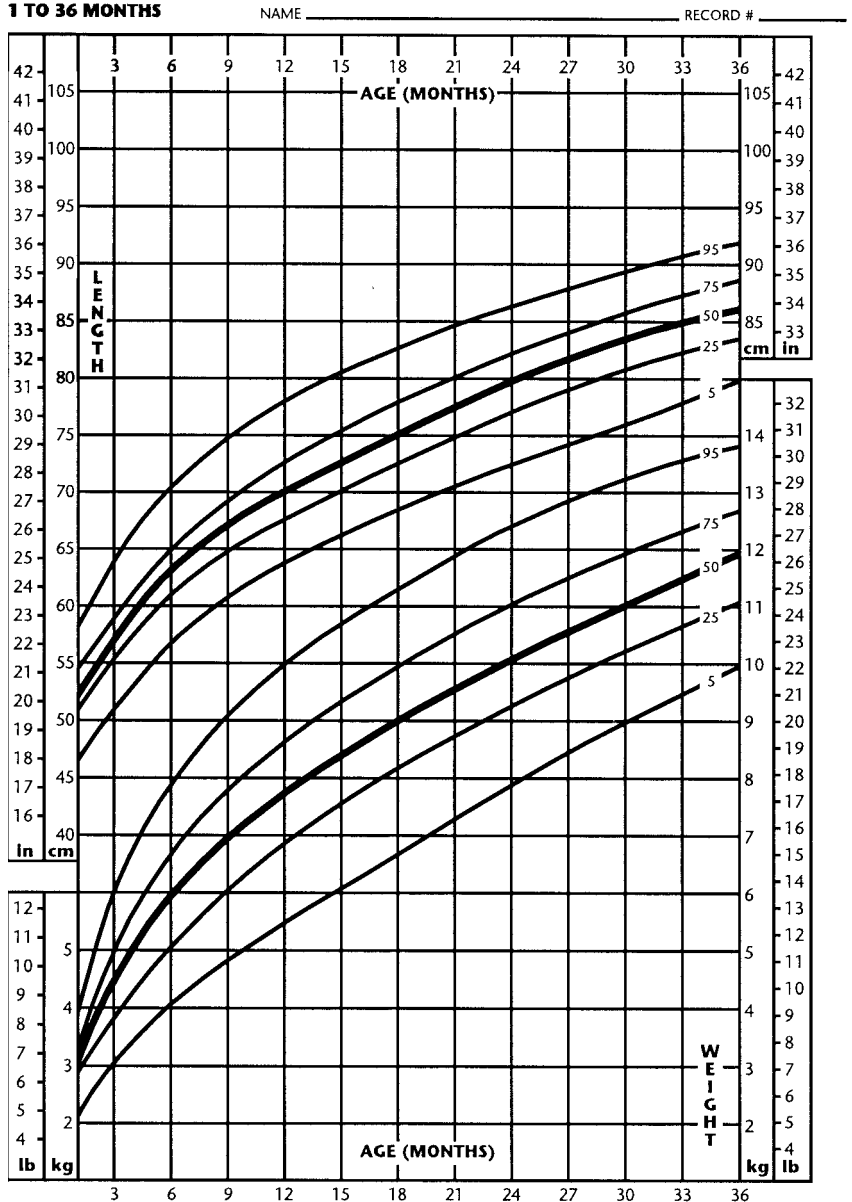
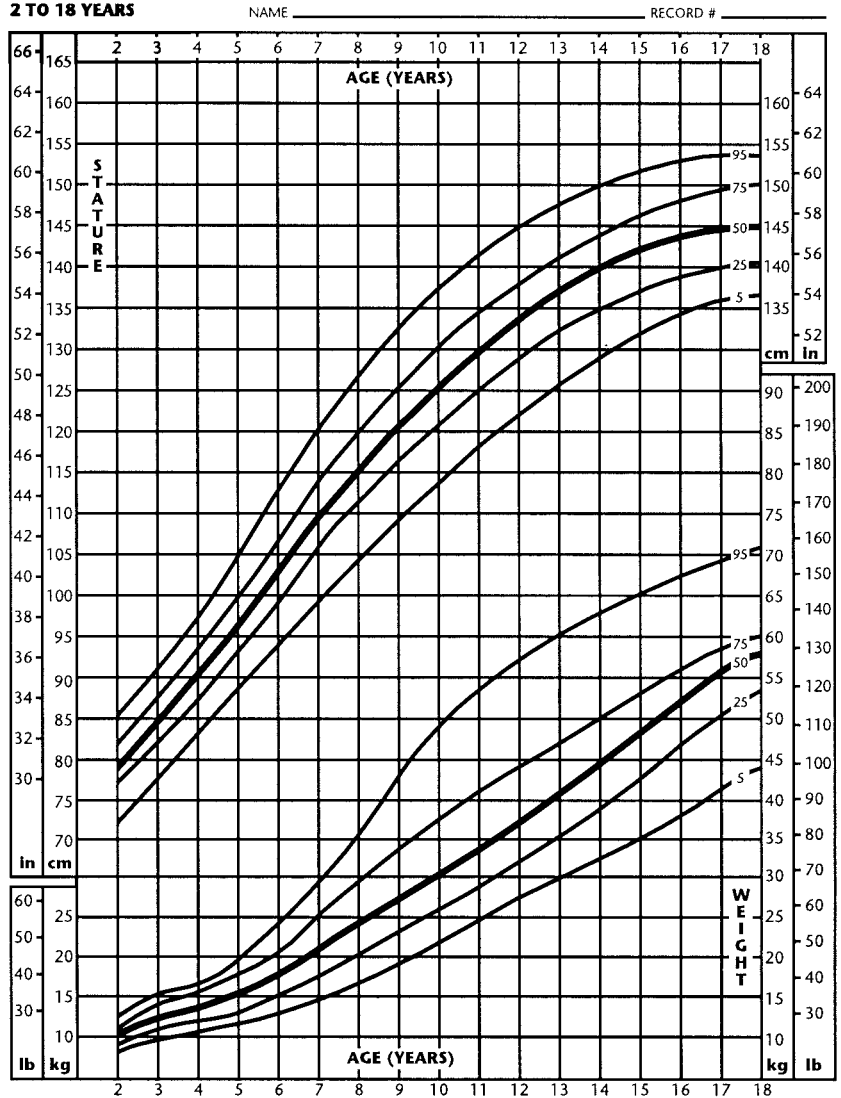


Fig 1. Percentiles for height and weight of females with Down syndrome, 1 to 36 months of age. From Cronk C, Crocker AC, Pueschel SM, et al. Growth charts for children with Down syndrome: 1 month to 18 years of age. *Pediatrics*. 1988;81:102–110.

**GIRLS WITH DOWN SYNDROME  
PHYSICAL GROWTH:  
2 TO 18 YEARS**



**Fig 2.** Percentiles for height and weight of females with Down syndrome, 2 to 18 years of age. From Cronk C, Crocker AC, Pueschel SM, et al. Growth charts for children with Down syndrome: 1 month to 18 years of age. *Pediatrics*. 1988;81:102–110.

(approximately 50%–70% risk of serous otitis media between 3 and 5 years).

- Check the child’s vision annually, using developmentally appropriate subjective and objective criteria. Refer the child to a pediatric ophthalmologist or ophthalmologist with special expertise and experience with children with disabilities every 2 years (approximately 50% risk of refractive errors between 3 and 5 years).
- At 3 to 5 years, obtain radiographs for evidence of atlantoaxial instability or subluxation. These may be obtained once during the preschool years. The need for these studies has been questioned, but they may be required for participation in the Special Olympics. These studies are more important for children who may participate in contact sports and are indicated in those who are symptomatic.<sup>16–19</sup>
- Perform thyroid screening tests annually.
- Discuss symptoms related to obstructive sleep apnea, including snoring, restless sleep, and sleep position. Refer to a specialist as indicated.<sup>20</sup>

**Anticipatory Guidance**

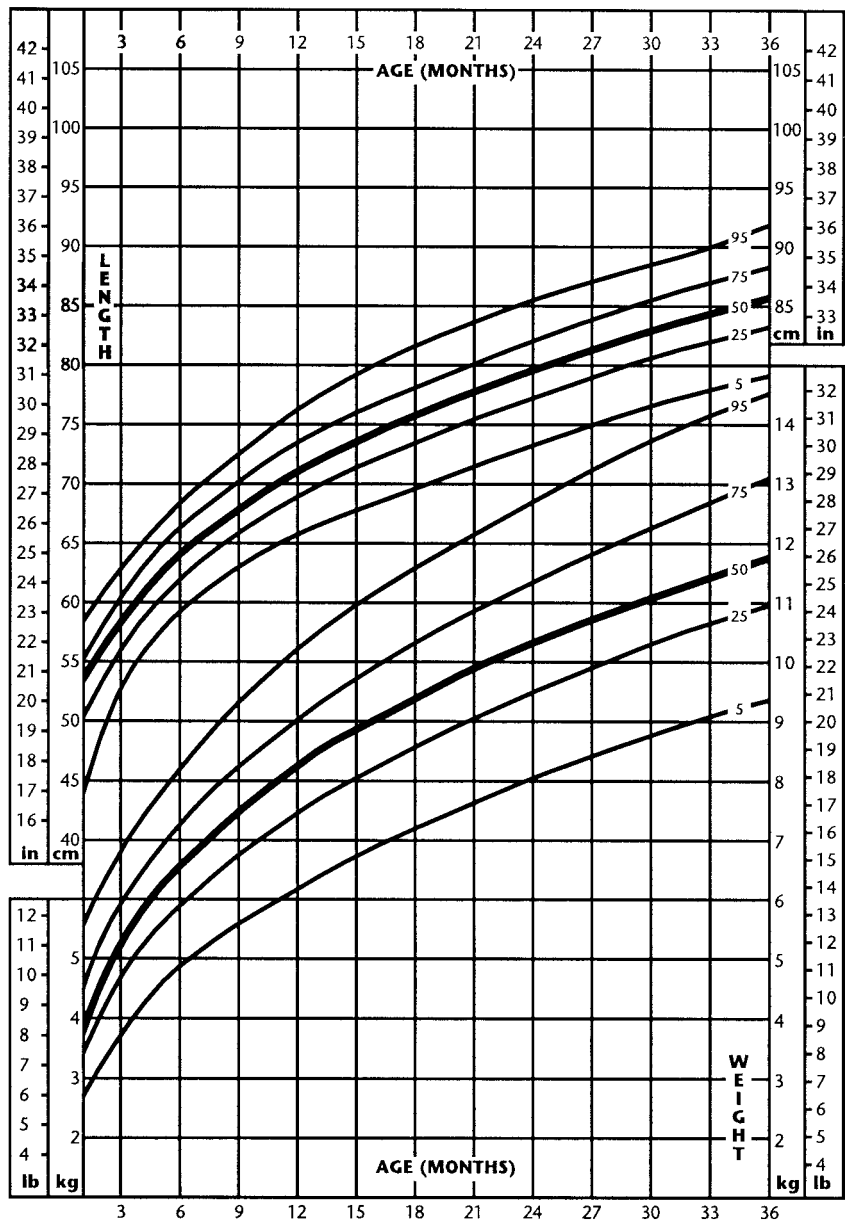
- Review early intervention, including physical therapy, occupational therapy, and speech, in the preschool program and discuss future school placement and performance.
- Discuss future pregnancy planning, risk of recurrence of Down syndrome, and prenatal diagnosis.
- Assess the child’s behavior, and talk about behavioral management, sibling adjustments, socialization, and recreational skills.
- Encourage families to establish optimal dietary and physical exercise patterns that will prevent obesity.

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

- Obtain a history and perform a physical examination with attention to growth and developmental status.
- Obtain audiologic evaluation annually.
- Obtain ophthalmologic evaluation annually.

**DOWN SYNDROME BOYS  
PHYSICAL GROWTH:  
1 TO 36 MONTHS**

NAME \_\_\_\_\_ RECORD # \_\_\_\_\_



**Fig 3.** Percentiles for height and weight of males with Down syndrome, 1 to 36 months of age. From Cronk C, Crocker AC, Pueschel SM, et al. Growth charts for children with Down syndrome: 1 month to 18 years of age. *Pediatrics*. 1988;81:102-110.

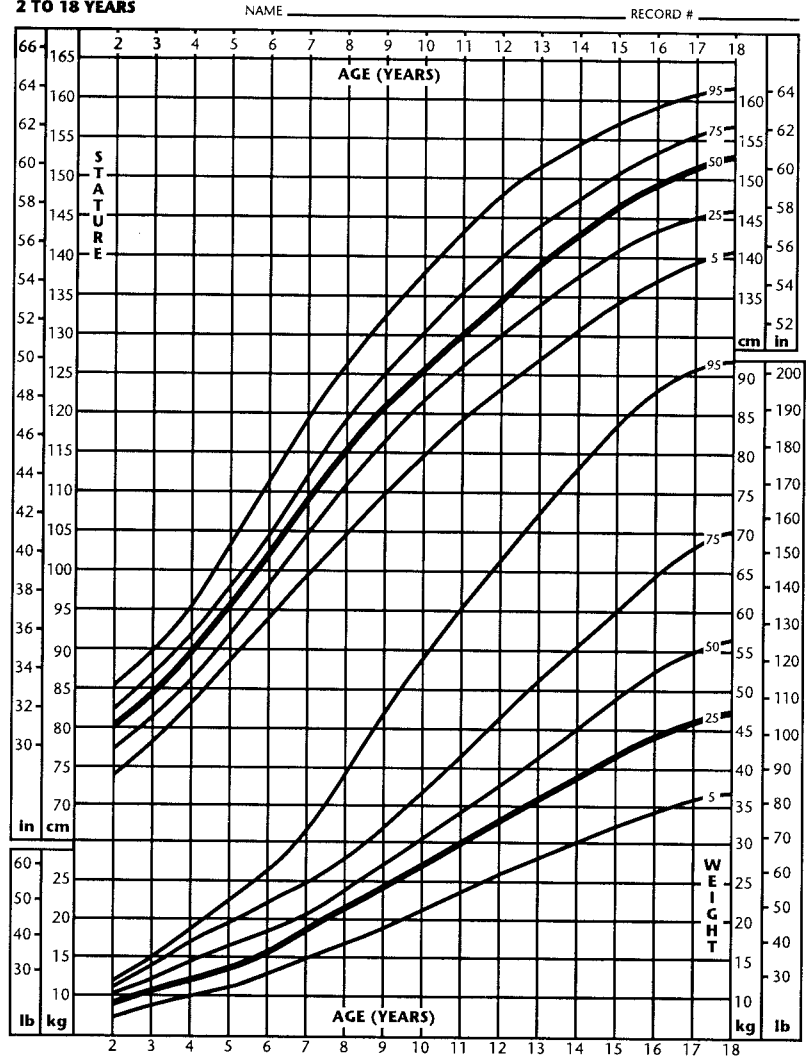
- Perform thyroid screening tests annually (3%–5% risk of hypothyroidism).
- If appropriate, discuss skin problems: very dry skin and other skin problems are particularly common in patients with Down syndrome.
- Discuss symptoms related to obstructive sleep apnea, including snoring, restless sleep, and sleep position. Refer to a specialist as indicated.<sup>20</sup>

**Anticipatory Guidance**

- Review the child’s development and appropriateness of school placement and developmental intervention.

- Discuss socialization, family status, and relationships, including financial arrangements and guardianship.
- Discuss the development of age-appropriate social skills, self-help skills, and the development of a sense of responsibility.
- Discuss psychosexual development, physical and sexual development, menstrual hygiene and management, fertility, and contraception.<sup>21</sup>
- Discuss the need for gynecologic care in the pubescent female. Talk about the recurrence risk of Down syndrome with the patient and her family if she were to become pregnant.<sup>22</sup> Review the fact that although there have been 2 case reports in

**BOYS WITH DOWN SYNDROME  
PHYSICAL GROWTH:  
2 TO 18 YEARS**



**Fig 4.** Percentiles for height and weight of males with Down syndrome, 2 to 18 years of age. From Cronk C, Crocker AC, Pueschel SM, et al. Growth charts for children with Down syndrome: 1 month to 18 years of age. *Pediatrics*. 1988;81:102-110.

which a male has reproduced, males with Down syndrome are usually infertile.<sup>22</sup>

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

**Examination**

- Perform physical examination including CBC and thyroid function tests.
- Obtain annual audiologic evaluation.
- Obtain annual ophthalmologic evaluation.
- Discuss skin care.

**Anticipatory Guidance**

- Discuss issues related to transition into adulthood.
- Discuss appropriateness of school placement with emphasis on adequate vocational training within the school curriculum.<sup>20,23</sup>
- Talk about the recurrence risk of Down syndrome with the patient and her family if she were to become pregnant.<sup>22</sup>

- Discuss sexuality and socialization. Discuss the need for and degree of supervision and/or the need for contraception. Make recommendations for routine gynecologic care.
- Discuss group homes and independent living opportunities, workshop settings, and other community-supported employment.
- Discuss intrafamily relationships, financial planning, and guardianship.
- Facilitate transfer to adult medical care.

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### Resources

- March of Dimes, Birth Defects Foundation, 1275 Mamaroneck Ave, White Plains, NY 10605; Telephone: 914/428-7100
- National Down Syndrome Congress, 1605 Chantilly Dr, Suite 250, Atlanta, GA 30324; Telephone: 404/633-1555 or 800/232-6372
- National Down Syndrome Society, 666 Broadway, New York, NY 10012; Telephone: 212/460-9330 or 800/221-4602