# Health Supervision for Children With Down Syndrome

Committee on Genetics

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 advice. Therefore, these guidelines offer advice for this situation as well.

Children with Down syndrome have multiple malformations and mental retardation due to 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, excess 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 retardation is variable, ranging from mild (IQ, 50 to 70) to moderate (IQ, 35 to 50), and only occasionally to severe (IQ, 20 to 35). There is an increased risk of congenital heart disease (50%); leukemia (<1%); deafness (75%); serous otitis media (50% to 70%); Hirschsprung disease (<1%); gastrointestinal atresias (12%); eye disease (60%), including cataracts (15%) and severe refractive errors (50%); acquired hip dislocation (6%); and thyroid disease (15%). Social quotient may be improved with early intervention techniques. Although level of function is exceedingly variable, children with Down syndrome often function better in social situations than might be expected from their IQ.

In about 95% of children with Down syndrome, the condition is due to nonfamilial trisomy 21. In approximately 3% to 4% of individuals with the Down syndrome phenotype, the extra chromosomal material is the result of an unbalanced translocation between chromosome 21 and another acrocentric chromosome. About three fourths of these unbalanced translocations are de novo, and about 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 individuals with the Down syndrome phenotype, two cell lines are present: one normal and one trisomy 21. This condition is called mosaicism. These individuals, on average, are affected less severely than individuals with trisomy 21 or translocated chromosome 21.

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 in caring for children with Down syndrome and their families.[1,2] It is organized by the issues that need to be addressed in the various age groups (see Table also).

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

- Review personal support available to family.
- Periodically review all the other financial and medical support programs for which the child and family may be eligible.
- Discuss filing for Supplemental Security Income (SSI) benefits.
- Discuss injury prevention with special consideration of developmental skills.
- Discuss 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 already have been provided for the family by a clinical geneticist and/or obstetrician. Because of a previous relationship with the family, however, the pediatrician may be called on to review this information and to assist in the decision-making process. As appropriate, the pediatrician should cover the following topics with the family:

1. Review and demonstrate the laboratory or imaging studies leading to the diagnosis.

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

| | Prenatal | Infancy, 1 mo–1 y | | | | | Early Childhood, 1–5 y | | | | | | Late Childhood, 5–13 y, Annual | Adolescence, 13–21 y, Annual |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | Neonatal | 2 mo | 4 mo | 6 mo | 9 mo | 12 mo | 15 mo | 18 mo | 24 mo | 3 y | 4 y | | |
| 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 | | ○ | ○ | ○ | ○ | ○ | ○ | ○ | ○ | ○ | ○ | ○ | ○ | ○ |
| Thyroid screening | | ○¶ | | ○ | | | ○ | | | ○ | ○ | ○ | ○ | ○ |
| Hearing screening | | | S/○ | S/○ | S/○ | S/○ | S/○‡ | | | S/○‡ | S/○‡ | S/○‡ | S/○§ | S/○ |
| Vision screening | | S/○ | S/○ | S/○ | S/○ | S/○‡ | S/○ | | | S/○ | S/○ | S/○ | S/○‖ | S/○ |
| Cervical spine roentgenogram | | | | | | | | | | | ○** | | | |
| Echocardiogram | ● | ○ | | | | | | | | | | | | |
| Psychosocial | | | | | | | | | | | | | | |
| Development and behavioral | S/○ | S/○ | S/○ | S/○ | S/○ | S/○ | S/○ | S/○ | S/○ | S/○ | S/○ | S/○ | S/○ | S/○ |
| School performance | | | | | | | | | | ○ | ○ | ○ | ○ | ○ |
| Socialization | | | | | | | S | | | | | | S | S |

* Assure compliance with the American Academy of Pediatrics "Recommendations for Preventive Pediatric Health Care."
● = to be performed; S = subjective, by history; and ○ = 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.

2. Explain the mechanism for occurrence of the disease in the fetus and the potential recurrence rate for the family.
3. Review the prognosis and manifestations, including any variability.
4. When applicable, recommend further studies that may refine the estimation of the prognosis (eg, fetal echocardiogram).
5. Review the currently available treatments and interventions. This discussion needs to include the efficacy, potential complications and/or side effects, costs, or other burdens of these treatments. Discuss any plausible future treatments.
6. Explore 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, adoption, etc.

If the pregnancy is continued, a plan for delivery and neonatal care must be developed with the obstetrician and the family. As the pregnancy progresses, further studies may be of value in 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 recurrence rates, future reproductive options, and evaluation of the risks of 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.

- Feeding problems.
- Hypotonia.
- Facial appearance.
- Check for strabismus, cataracts, and nystagmus at birth or by 6 months.
- Heart defects (~50% risk). Perform cardiac evaluation (echocardiogram recommended).
- Duodenal atresia.
- Leukemia—more common in children with Down syndrome than in the general population, but still rare (<1%); leukemoid reactions, on the other hand, are common.
- Congenital hypothyroidism (1% risk).
- Increased susceptibility to respiratory tract infections.

### Anticipatory Guidance

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 prenatal diagnosis and recurrence risk in subsequent pregnancies.

- Trisomy 21 has a recurrence risk of 1 in 100 until maternal age 35, when age-determined risks take precedence. Other family members do not have an increased risk of bearing children with abnormal chromosomes.
- Recurrence risk assessment in translocations is more complex; the pediatrician should consult a genetics specialist in such cases.

Discuss unproven therapies.[3]

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

### Examination

Physical examination and laboratory studies.

- Monitor the infant's hearing at each health supervision visit and review the risk of serous otitis media (~50% to 70%).[4] Use developmentally appropriate subjective and objective criteria. Refer the infant to an audiologist if necessary.
- Check for strabismus, cataracts, and nystagmus by 6 months, if not done at birth. At 6 to 12 months, check the infant's vision at each visit, using developmentally appropriate subjective and objective criteria. Refer the infant to an ophthalmologist if necessary by 9 months.
- Perform thyroid screening tests. Repeat at 4 to 6 months and at 12 months.[5]

### Anticipatory Guidance

- Review the infant's growth and development relative to other children with Down syndrome (see Down syndrome Growth Developmental Assessment Charts).[6]
- Review availability of Down syndrome support groups. (See "Bibliography and Resources for New Parents.")
- Observe 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.") Check status at 6 to 12 months.

- Review the risk of recurrence of Down syndrome and prenatal diagnosis during the first year.

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

### Examination

Physical examination and laboratory studies.

- Check the child's hearing annually, using developmentally appropriate subjective and objective criteria. Refer the child to an audiologist if necessary (~50% to 70% risk of serous otitis between 3 and 5 years).
- Check the child's vision annually, using developmentally appropriate subjective and objective criteria. Refer the child to an ophthalmologist if necessary (~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 in 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 in individuals who may participate in contact sports and are indicated in those who are symptomatic.[7–10]
- Perform thyroid screening tests annually.

### Anticipatory Guidance

- Review 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.
- At 3 to 5 years, discuss advantages and disadvantages of plastic surgery for facial appearance and speech.[11]

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

### Examination

Physical examination and laboratory studies.

- Perform audiologic evaluation at least once during this time frame.
- Perform ophthalmologic evaluation as needed.
- Perform annual thyroid screening tests (3% to 5% risk).
- If appropriate, discuss skin problems: very dry skin and other skin problems are particularly common in patients with Down syndrome.

### Anticipatory Guidance

- Review the development and appropriateness of school placement with emphasis on prevocational skills.
- Discuss socialization and family status and relationships, including financial arrangements and guardianship.
- Discuss the development of age-appropriate social skills and the development of a sense of responsibility.
- Discuss psychosexual development, physical sexual development, menstrual hygiene and management, and fertility in both girls and boys.[12]
- Emphasize socialization skills. Discuss contraception and make recommendations. Talk about the recurrence risk to the patient and her family if she were to become pregnant.[13] (Although there has been one case report in which a male has reproduced, males with Down syndrome are usually infertile.)

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

### Examination

Physical examination and laboratory studies.

- Perform annual audiologic evaluation.
- Perform annual ophthalmologic evaluation.
- Perform annual thyroid screening tests.
- 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.[14]
- Discuss sexuality and socialization. Discuss the need for and degree of supervision and/or the need for contraception. Make recommendations.
- Discuss group homes, workshop settings, and other community-supported employment.
- Discuss intrafamilial relationships, financial planning, and guardianship.
- Facilitate transfer to adult medical care, if appropriate or desired.



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National Down Syndrome Congress, 1605 Chantilly Dr, Suite 250, Atlanta, GA 30324. Tel: 404/633-1555 or 800/232-6372.

National Down Syndrome Society, 666 Broadway, New York, NY 10012. Tel: 212/460-9330 or 800/221-4602.

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